Contents

Module 7 Infectious disease

Module 8 Non-infectious disease and disorders

How to use this book

Pearson Biology 12 New South Wales

Pearson Biology 12 New South Wales has been written to fully align with the new Stage 6 Syllabus for New South Wales Biology. The book covers Modules 5 to 8 in an easy-to-use resource. Explore how to use this book below.

Chapter opener

The chapter opening page links the syllabus to the chapter content. Key content addressed in the chapter is clearly listed.


CHAPTER 04

DNA and polypeptide synthesis

By the end of this chapter you will be able to describe the differences and similarities of the forms in which DNA exists in eukaryotic and prokaryotic cells. You will also understand the role of nucleic acids (DNA and multiple forms of RNA) as molecules that encode instructions for polypeptide synthesis from amino acids. You will also examine the process of polypeptide synthesis from transcription to translation.

The complexity and importance of proteins will become evident, starting with their construction from polypeptides to the many functions they play in living organisms. You will learn that nearly every function of a living organism depends on proteins.

Content

INQUIRY QUESTION

Why is polypeptide synthesis important?

By the end of this chapter you will be able to:

- construct appropriate representations to model and compare the forms in which DNA exists in eukaryotes and prokaryotes (ACSBL076) CE1
- model the process of polypeptide synthesis, including: (ACSBL079)
  - transcription and translation
  - assessing the importance of mRNA and tRNA in transcription and translation (ACSBL079)
  - analysing the function and importance of polypeptide synthesis (ACSBL080)
  - assessing how genes and environment affect phenotypic expression (ACSBL081) CE1 L
- investigate the structure and function of proteins in living things L

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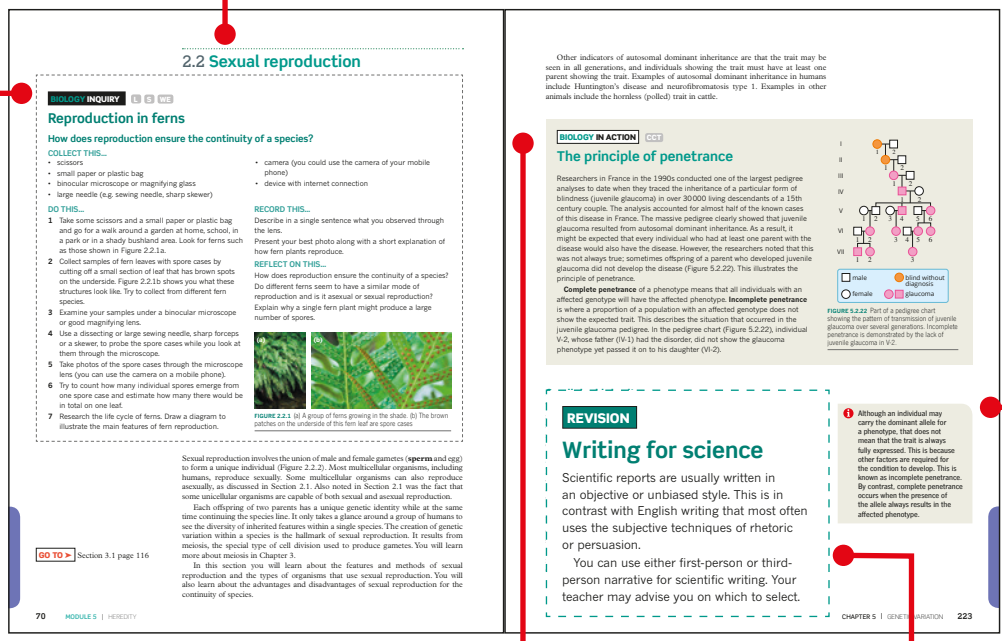
Section

Each chapter is clearly divided into manageable sections of work. Best-practice literacy and instructional design are combined with high-quality, relevant photos and illustrations to help students better understand the ideas or concepts being developed.


2.2 Sexual reproduction

BIOLOGY INQUIRY L S WE

Reproduction in ferns

How does reproduction ensure the continuity of a species?

COLLECT THIS...

- scissors
- small paper or plastic bag
- binocular microscope or magnifying glass
- large needle (e.g. sewing needle, sharp skewer)
- camera (you could use the camera of your mobile phone)
- device with internet connection

DO THIS...

1. Take some scissors and a small paper or plastic bag and go for a walk around a garden at home, school, in a park or in a shady bushland area. Look for ferns such as those shown in Figure 2.2.1a.
2. Collect samples of fern leaves with spore cases by cutting off a small section of leaf that has brown spots on the underside. Figure 2.2.1b shows you what these structures look like. Try to collect from different fern species.
3. Examine your samples under a binocular microscope or good magnifying lens.
4. Use a dissecting or large sewing needle, sharp forceps or a skewer, to probe the spore cases while you look at them through the microscope.
5. Take photos of the spore cases through the microscope lens (you can use the camera on a mobile phone).
6. Try to count how many individual spores emerge from one spore case and estimate how many there would be in total on one leaf.
7. Research the life cycle of ferns. Draw a diagram to illustrate the main features of fern reproduction.

RECORD THIS...

Describe in a single sentence what you observed through the lens.

Present your best photo along with a short explanation of how fern plants reproduce.

REFLECT ON THIS...

How does reproduction ensure the continuity of a species?

Do different ferns seem to have a similar mode of reproduction and is it asexual or sexual reproduction?

Explain why a single fern plant might produce a large number of spores.

FIGURE 2.2.1 (a) A group of ferns growing in the shade. (b) The brown patches on the underside of this fern leaf are spore cases.

Sexual reproduction involves the union of male and female gametes (**sperm** and egg) to form a unique individual (Figure 2.2.2). Most multicellular organisms, including humans, reproduce sexually. Some multicellular organisms can also reproduce asexually, as discussed in Section 2.1. Also noted in Section 2.1 was the fact that some unicellular organisms are capable of both sexual and asexual reproduction.

Each offspring of two parents has a unique genetic identity while at the same time continuing the species line. It only takes a glance around a group of humans to see the diversity of inherited features within a single species. The creation of genetic variation within a species is the hallmark of sexual reproduction. It results from meiosis, the special type of cell division used to produce gametes. You will learn more about meiosis in Chapter 3.

In this section you will learn about the features and methods of sexual reproduction and the types of organisms that use sexual reproduction. You will also learn about the advantages and disadvantages of sexual reproduction for the continuity of species.

GO TO > Section 3.1 page 116

70 MODULE 5 | HEREDITY

Other indicators of autosomal dominant inheritance are that the trait may be seen in all generations, and individuals showing the trait must have at least one parent showing the trait. Examples of autosomal dominant inheritance in humans include Huntington's disease and neurofibromatosis type 1. Examples in other animals include the hornless (polled) trait in cattle.

BIOLOGY IN ACTION CC1

The principle of penetrance

Researchers in France in the 1990s conducted one of the largest pedigree analyses to date when they traced the inheritance of a particular form of blindness (juvenile glaucoma) in over 30000 living descendants of a 15th century couple. The analysis accounted for almost half of the known cases of this disease in France. The massive pedigree clearly showed that juvenile glaucoma resulted from autosomal dominant inheritance. As a result, it might be expected that every individual who had at least one parent with the disease would also have the disease. However, the researchers noted that this was not always true; sometimes offspring of a parent who developed juvenile glaucoma did not develop the disease (Figure 5.2.22). This illustrates the principle of penetrance.

Complete penetrance of a phenotype means that all individuals with an affected genotype will have the affected phenotype. **Incomplete penetrance** is where a proportion of a population with an affected genotype does not show the expected trait. This describes the situation that occurred in the juvenile glaucoma pedigree. In the pedigree chart (Figure 5.2.22), individual V-2, whose father (IV-1) had the disorder, did not show the glaucoma phenotype yet passed it on to his daughter (VI-2).

male
female
blind without diagnosis
glaucoma

FIGURE 5.2.22 Part of a pedigree chart showing the pattern of transmission of juvenile glaucoma over several generations. Incomplete penetrance is demonstrated by the lack of juvenile glaucoma in V-2.

REVISION

Writing for science

Scientific reports are usually written in an objective or unbiased style. This is in contrast with English writing that most often uses the subjective techniques of rhetoric or persuasion.

You can use either first-person or third-person narrative for scientific writing. Your teacher may advise you on which to select.

Although an individual may carry the dominant allele for a phenotype, that does not mean that the trait is always fully expressed. This is because other factors are required for the condition to develop. This is known as incomplete penetrance. By contrast, complete penetrance occurs when the presence of the allele always results in the affected phenotype.

CHAPTER 5 | GENETIC VARIATION 223

Highlight box

Highlight boxes focus students' attention on important information such as key definitions and summary points.

Biology Inquiry

Biology Inquiry features are inquiry-based activities that assist students to discover concepts before learning about them. They encourage students to think about what happens in the world and how science can provide explanations.

Biology in Action

Biology in Action boxes place biology in an applied situation or a relevant context. These refer to the nature and practice of biology, its applications and associated issues, and the historical development of its concepts and ideas.

Revision box

Revision boxes are used to remind students of vital concepts previously covered that are required for current learning.

BioFile

BioFiles include a range of interesting and real-world examples to engage students.

SkillBuilder

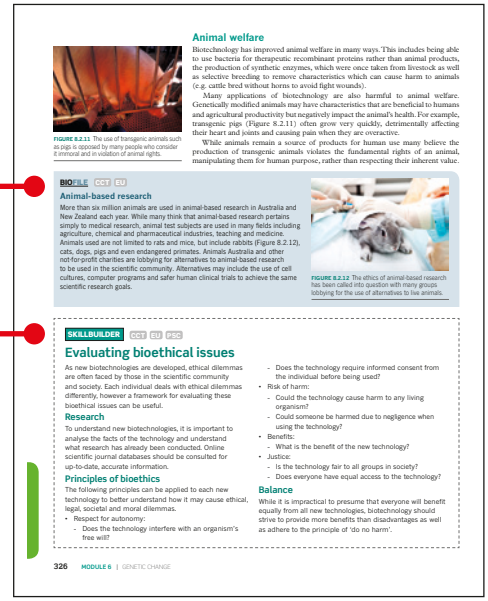
A SkillBuilder outlines a method or technique. They are instructive and self-contained. They step students through the skill to support science application.

Worked examples

Worked examples are set out in steps that show thinking and working. This format greatly enhances student understanding by clearly linking underlying logic to the relevant calculations. Each Worked example is followed by a Try yourself activity. This mirror problem allows students to immediately test their understanding.

Additional content

Additional content includes material that goes beyond the core content of the syllabus. They are intended for students who wish to expand their depth of understanding in a particular area.



Animal welfare

Biotechnology has improved animal welfare in many ways. This includes being able to use bacteria for therapeutic recombinant proteins rather than animal products, the production of synthetic enzymes, which were once taken from livestock as well as selective breeding to remove characteristics which can cause harm to animals (e.g. cattle bred without horns to avoid fight wounds).

Many applications of biotechnology are also harmful to animal welfare. Genetically modified animals may have characteristics that are beneficial to humans and agricultural productivity but negatively impact the animal's health. For example, transgenic pigs (Figure 8.2.11) often grow very quickly, detrimentally affecting their heart and joints and causing pain when they are overactive.

While animals remain a source of products for human use many believe the production of transgenic animals violates the fundamental rights of an animal, manipulating them for human purpose, rather than respecting their inherent value.

FIGURE 8.2.11 The use of transgenic animals such as pigs is opposed by many people who consider it immoral and in violation of animal rights.

BIOFILE CCT EU

Animal-based research

More than six million animals are used in animal-based research in Australia and New Zealand each year. While many think that animal-based research pertains simply to medical research, animal test subjects are used in many fields including agriculture, chemical and pharmaceutical industries, teaching and medicine. Animals used are not limited to rats and mice, but include rabbits (Figure 8.2.12), cats, dogs, pigs and even endangered primates. Animals Australia and other not-for-profit charities are lobbying for alternatives to animal-based research to be used in the scientific community. Alternatives may include the use of cell cultures, computer programs and safer human clinical trials to achieve the same scientific research goals.

FIGURE 8.2.12 The ethics of animal-based research has been called into question with many groups lobbying for the use of alternatives to live animals.

SKILLBUILDER CCT EU PEC

Evaluating bioethical issues

As new biotechnologies are developed, ethical dilemmas are often faced by those in the scientific community and society. Each individual deals with ethical dilemmas differently, however a framework for evaluating these bioethical issues can be useful.

Research

To understand new biotechnologies, it is important to analyse the facts of the technology and understand what research has already been conducted. Online scientific journal databases should be consulted for up-to-date, accurate information.

Principles of bioethics

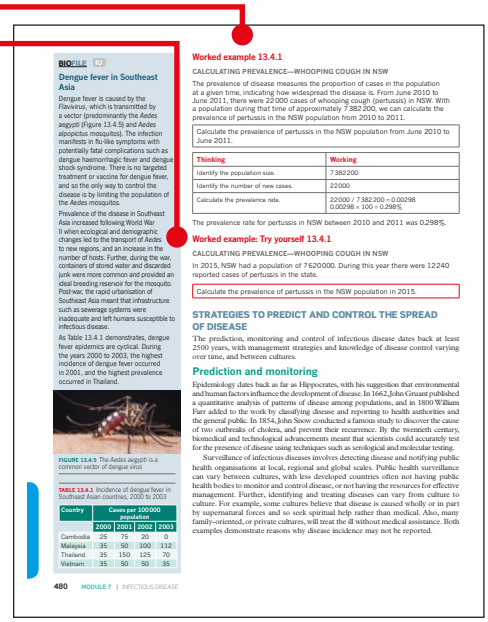
The following principles can be applied to each new technology to better understand how it may cause ethical, legal, societal and moral dilemmas.

- Respect for autonomy:
  - Does the technology interfere with an organism's free will?
  - Does the technology require informed consent from the individual before being used?
- Risk of harm:
  - Could the technology cause harm to any living organism?
  - Could someone be harmed due to negligence when using the technology?
- Benefits:
  - What is the benefit of the new technology?
- Justice:
  - Is the technology fair to all groups in society?
  - Does everyone have equal access to the technology?

Balance

While it is impractical to presume that everyone will benefit equally from all new technologies, biotechnology should strive to provide more benefits than disadvantages as well as adhere to the principle of 'do no harm'.

326 MODULE 6 | GENETIC CHANGE



BIOFILE EU

Dengue fever in Southeast Asia

Dengue fever is caused by the *Flavivirus*, which is transmitted by a vector (predominantly the *Aedes aegypti* (Figure 13.4.5) and *Aedes albopictus* mosquitos). The infection manifests in flu-like symptoms with potentially fatal complications such as dengue haemorrhagic fever and dengue shock syndrome. There is no targeted treatment or vaccine for dengue fever, and so the only way to control the disease is by limiting the population of the *Aedes* mosquitos.

Prevalence of the disease in Southeast Asia increased following World War II when ecological and demographic changes led to the transport of *Aedes* to new regions, and an increase in the number of hosts. Further, during the war, containers of stored water and discarded junk were more common and provided an ideal breeding reservoir for the mosquito. Post-war, the rapid urbanisation of Southeast Asia meant that infrastructure such as sewerage systems were inadequate and left humans susceptible to infectious disease.

As Table 13.4.1 demonstrates, dengue fever epidemics are cyclical. During the years 2000 to 2003, the highest incidence of dengue fever occurred in 2001, and the highest prevalence occurred in Thailand.

FIGURE 13.4.5 The *Aedes aegypti* is a common vector of dengue virus

TABLE 13.4.1 Incidence of dengue fever in Southeast Asian countries, 2000 to 2003

Country	Cases per 100 000 population			
	2000	2001	2002	2003
Cambodia	25	75	20	0
Malaysia	35	50	100	112
Thailand	35	150	125	70
Vietnam	35	50	50	35

Worked example 13.4.1

CALCULATING PREVALENCE—WHOOPING COUGH IN NSW

The prevalence of disease measures the proportion of cases in the population at a given time, indicating how widespread the disease is. From June 2010 to June 2011, there were 22 000 cases of whooping cough (pertussis) in NSW. With a population during that time of approximately 7 382 200, we can calculate the prevalence of pertussis in the NSW population from 2010 to 2011.

Calculate the prevalence of pertussis in the NSW population from June 2010 to June 2011.

Thinking	Working
Identify the population size.	7 382 200
Identify the number of new cases.	22 000
Calculate the prevalence rate.	22 000 / 7 382 200 = 0.00298 0.00298 × 100 = 0.298%

The prevalence rate for pertussis in NSW between 2010 and 2011 was 0.298%.

Worked example: Try yourself 13.4.1

CALCULATING PREVALENCE—WHOOPING COUGH IN NSW

In 2015, NSW had a population of 7 620 000. During this year there were 12 240 reported cases of pertussis in the state.

Calculate the prevalence of pertussis in the NSW population in 2015.

STRATEGIES TO PREDICT AND CONTROL THE SPREAD OF DISEASE

The prediction, monitoring and control of infectious disease dates back at least 2500 years, with management strategies and knowledge of disease control varying over time, and between cultures.

Prediction and monitoring

Epidemiology dates back as far as Hippocrates, with his suggestion that environmental and human factors influence the development of disease. In 1662, John Graunt published a quantitative analysis of patterns of disease among populations, and in 1800 William Farr added to the work by classifying disease and reporting to health authorities and the general public. In 1854, John Snow conducted a famous study to discover the cause of two outbreaks of cholera, and prevent their recurrence. By the twentieth century, biomedical and technological advancements meant that scientists could accurately test for the presence of disease using techniques such as serological and molecular testing.

Surveillance of infectious diseases involves detecting disease and notifying public health organisations at local, regional and global scales. Public health surveillance can vary between cultures, with less developed countries often not having public health bodies to monitor and control disease, or not having the resources for effective management. Further, identifying and treating diseases can vary from culture to culture. For example, some cultures believe that disease is caused wholly or in part by supernatural forces and so seek spiritual help rather than medical. Also, many family-oriented, or private cultures, will treat the ill without medical assistance. Both examples demonstrate reasons why disease incidence may not be reported.

480 MODULE 7 | INFECTIOUS DISEASE

+ ADDITIONAL

Hybridisation and artificial pollination

Gregor Mendel (Figure 2.4.22), often called the father of modern genetics, investigated hereditary patterns of hybrids. Hybrids are the offspring from two organisms that are of different breeds, varieties or species. Hybrids create new cultivars, from new apple varieties to tangelos to hybrid corn. Some mammals produce hybrids; a mule is the progeny of a horse and a donkey. Crossbreeding of stock has produced the highly rated Brahman cattle breed and hybrids from it that are credited with saving the cattle industry in Australia (Figure 2.4.23). Hybridisation is in fact a form of selective breeding and is widely practiced by home gardeners, horticulturists, farmers and research scientists.

Mendel chose common garden pea plants (*Pisum sativum*) for the focus of his research because they can be grown easily in large numbers and their reproduction can be manipulated. In his experiments, Mendel was able to selectively cross-pollinate pumbred plants with particular traits and observe the outcome over many generations. This was the basis for his conclusions about the nature of genetic inheritance. Mendel first had to make sure the flowers did not pollinate themselves. To do that, he used forceps to remove the anthers, their pollen-bearing parts. Then he immediately dusted pollen from other selected pea plants, using a fine paintbrush, onto the stigma of the altered plants to fertilise them. In this way he was able to control the breeding of the pea plants.

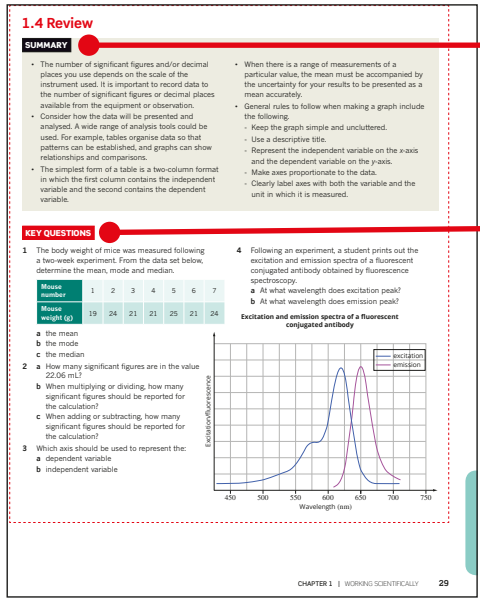
Many home gardeners practice selective pollination by hand (Figure 2.4.24). As a general rule, they transfer pollen in the early morning while it is likely to be most healthy. A simple technique is to touch a cotton bud or brush tip to the pollen-covered anther on the chosen flower. Alternatively, tweezers can be used to pluck off the anther and filament and use the entire structure for pollination. They then touch the pollen-covered cotton bud, brush or the anther itself, to the receptive stigma (look for a shiny, sticky surface) on the chosen flower. A successful attempt will see pollen grains adhering to the stigma.

FIGURE 2.4.22 Gregor Mendel (1822–1884), Austrian monk and scientist, who founded the field of genetics with his experiments involving selective breeding of pea plants.

FIGURE 2.4.23 Brahman cattle, first crossbred in the USA from four different Indian zebu breeds, have been hugely successful in the harsh Australian climate and because of their tick resistance. Over 50% of Australian cattle are now either Brahman, or Brahman crossed with European breeds.

FIGURE 2.4.24 A home gardener pollinating cucumber blossoms by hand with a cotton bud

CHAPTER 2 | REPRODUCTION 109



1.4 Review

SUMMARY

- The number of significant figures and/or decimal places you use depends on the scale of the instrument used. It is important to record data to the number of significant figures or decimal places available from the equipment or observation.
- Consider how the data will be presented and analysed. A wide range of analysis tools could be used. For example, tables organise data so that patterns can be established, and graphs can show relationships and comparisons.
- The simplest form of a table is a two-column format in which the first column contains the independent variable and the second contains the dependent variable.
- When there is a range of measurements of a particular value, the mean must be accompanied by the uncertainty for your results to be presented as a mean accurately.
- General rules to follow when making a graph include the following.
  - Keep the graph simple and uncluttered.
  - Use a descriptive title.
  - Represent the independent variable on the *x*-axis and the dependent variable on the *y*-axis.
  - Make axes proportionate to the data.
  - Clearly label axes with both the variable and the unit in which it is measured.

KEY QUESTIONS

1 The body weight of mice was measured following a two-week experiment. From the data set below, determine the mean, mode and median.

Mouse number	1	2	3	4	5	6	7
Mouse weight (g)	19	24	21	21	25	21	24

a the mean
b the mode
c the median

2 a How many significant figures are in the value 22.06 mL?
b When multiplying or dividing, how many significant figures should be reported for the calculation?
c When adding or subtracting, how many significant figures should be reported for the calculation?

3 Which axis should be used to represent the:
a dependent variable
b independent variable

4 Following an experiment, a student prints out the excitation and emission spectra of a fluorescent conjugated antibody obtained by fluorescence spectroscopy.
a At what wavelength does excitation peak?
b At what wavelength does emission peak?

Excitation and emission spectra of a fluorescent conjugated antibody

CHAPTER 1 | WORKING SCIENTIFICALLY 29

Section summary

Each section has a summary to help students consolidate the key points and concepts.

Section review questions

Each section finishes with key questions to test students' understanding and ability to recall the key concepts of the section.

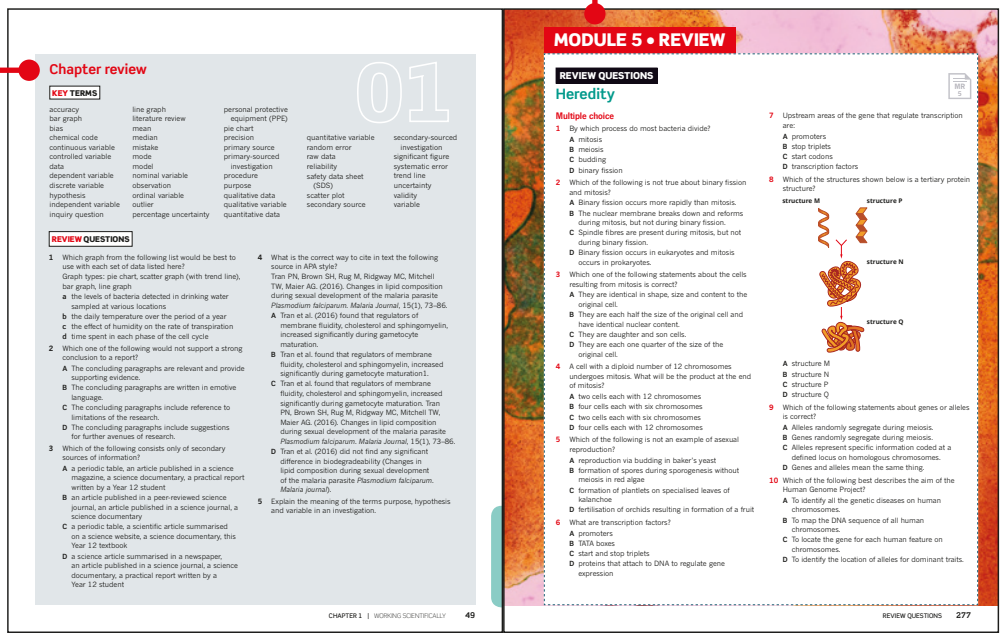
How to use this book

Chapter review

Each chapter finishes with a list of key terms covered in the chapter and a set of questions to test students' ability to apply the knowledge gained from the chapter.

Module review

Each module finishes with a comprehensive set of questions, including multiple choice and short answer. These assist students in drawing together their knowledge and understanding, and applying it to these types of questions.



Chapter review

01

KEY TERMS

accuracy
bar graph
bias
chemical code
continuous variable
controlled variable
data
dependent variable
discrete variable
hypothesis
independent variable
inquiry question
line graph
literature review
mean
median
mistake
mode
model
nominal variable
observation
ordinal variable
outlier
percentage uncertainty
personal protective equipment (PPE)
pie chart
precision
primary source
primary-sourced investigation
procedure
purpose
qualitative data
qualitative variable
quantitative data
quantitative variable
random error
raw data
reliability
safety data sheet (SDS)
scatter plot
secondary source
secondary-sourced investigation
significant figure
systematic error
trend line
uncertainty
validity
variable

REVIEW QUESTIONS

1 Which graph from the following list would be best to use with each set of data listed here?
Graph types: pie chart, scatter graph (with trend line), bar graph, line graph
a the levels of bacteria detected in drinking water sampled at various locations
b the daily temperature over the period of a year
c the effect of humidity on the rate of transpiration
d time spent in each phase of the cell cycle

2 Which one of the following would not support a strong conclusion to a report?
A The concluding paragraphs are relevant and provide supporting evidence.
B The concluding paragraphs are written in emotive language.
C The concluding paragraphs include reference to limitations of the research.
D The concluding paragraphs include suggestions for further avenues of research.

3 Which of the following consists only of secondary sources of information?
A a periodic table, an article published in a science magazine, a science documentary, a practical report written by a Year 12 student
B an article published in a peer-reviewed science journal, an article published in a science journal, a science documentary
C a periodic table, a scientific article summarised on a science website, a science documentary, this Year 12 textbook
D a science article summarised in a newspaper, an article published in a science journal, a science documentary, a practical report written by a Year 12 student

4 What is the correct way to cite in text the following source in APA style?
Tran PN, Brown SH, Rug M, Ridgway MC, Mitchell TW, Maier AG. (2016). Changes in lipid composition during sexual development of the malaria parasite *Plasmodium falciparum*. *Malaria Journal*, 15(1), 73–86.
A Tran et al. (2016) found that regulators of membrane fluidity, cholesterol and sphingomyelin, increased significantly during gametocyte maturation.
B Tran et al. found that regulators of membrane fluidity, cholesterol and sphingomyelin, increased significantly during gametocyte maturation1.
C Tran et al. found that regulators of membrane fluidity, cholesterol and sphingomyelin, increased significantly during gametocyte maturation. Tran PN, Brown SH, Rug M, Ridgway MC, Mitchell TW, Maier AG. (2016). Changes in lipid composition during sexual development of the malaria parasite *Plasmodium falciparum*. *Malaria Journal*, 15(1), 73-86.
D Tran et al. (2016) did not find any significant difference in biodegradeability (Changes in lipid composition during sexual development of the malaria parasite *Plasmodium falciparum*. *Malaria journal*).

5 Explain the meaning of the terms purpose, hypothesis and variable in an investigation.

CHAPTER 1 | WORKING SCIENTIFICALLY 49

MODULE 5 • REVIEW

REVIEW QUESTIONS

Heredity

Multiple choice

1 By which process do most bacteria divide?
A mitosis
B meiosis
C budding
D binary fission

2 Which of the following is not true about binary fission and mitosis?
A Binary fission occurs more rapidly than mitosis.
B The nuclear membrane breaks down and reforms during mitosis, but not during binary fission.
C Spindle fibres are present during mitosis, but not during binary fission.
D Binary fission occurs in eukaryotes and mitosis occurs in prokaryotes.

3 Which one of the following statements about the cells resulting from mitosis is correct?
A They are identical in shape, size and content to the original cell.
B They are each half the size of the original cell and have identical nuclear content.
C They are daughter and son cells.
D They are each one quarter of the size of the original cell.

4 A cell with a diploid number of 12 chromosomes undergoes mitosis. What will be the product at the end of mitosis?
A two cells each with 12 chromosomes
B four cells each with six chromosomes
C two cells each with six chromosomes
D four cells each with 12 chromosomes

5 Which of the following is not an example of asexual reproduction?
A reproduction via budding in baker's yeast
B formation of spores during sporogenesis without meiosis in red algae
C formation of plantlets on specialised leaves of kalanchoe
D fertilisation of orchids resulting in formation of a fruit

6 What are transcription factors?
A promoters
B TATA boxes
C start and stop triplets
D proteins that attach to DNA to regulate gene expression

7 Upstream areas of the gene that regulate transcription are:
A promoters
B stop triplets
C start codons
D transcription factors

8 Which of the structures shown below is a tertiary protein structure?
structure M
structure P
structure N
structure Q
A structure M
B structure N
C structure P
D structure Q

9 Which of the following statements about genes or alleles is correct?
A Alleles randomly segregate during meiosis.
B Genes randomly segregate during meiosis.
C Alleles represent specific information coded at a defined locus on homologous chromosomes.
D Genes and alleles mean the same thing.

10 Which of the following best describes the aim of the Human Genome Project?
A To identify all the genetic diseases on human chromosomes.
B To map the DNA sequence of all human chromosomes.
C To locate the gene for each human feature on chromosomes.
D To identify the location of alleles for dominant traits.

REVIEW QUESTIONS 277

Icons

The New South Wales Stage 6 syllabus 'Learning across the curriculum' and 'General capabilities' content are addressed throughout the series and are identified using the following icons.

AHC A CC CCT DD EU ICT
IU L N PSC S WE

'Go to' icons are used to make important links to relevant content within the same Student Book.

GO TO ➤

This icon indicates the best time to engage with a worksheet (WS), a practical activity (PA), a depth study (DS) or module review (MR) questions in *Pearson Biology 12 New South Wales Skills and Assessment* book.

WS 3.1

This icon indicates the best time to engage with a practical activity on *Pearson Biology 12 New South Wales* Reader+.

PA 3.2

Glossary

Key terms are shown in **bold** in sections and listed at the end of each chapter. A comprehensive glossary at the end of the book includes and defines all the key terms.

Answers

Comprehensive answers and fully worked solutions for all section review questions, Worked example: Try yourself features, chapter review questions and module review questions are provided via *Pearson Biology 12 New South Wales* Reader+.

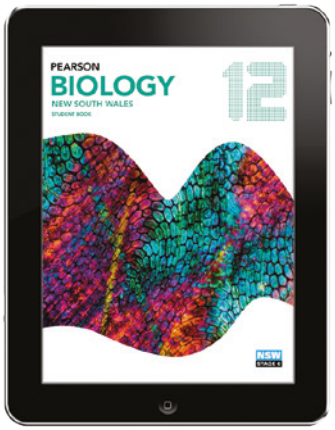
Pearson Biology 12
New South Wales

Student Book

Pearson Biology 12 New South Wales has been written to fully align with the new Stage 6 syllabus for New South Wales. The Student Book includes the very latest developments and applications of biology and incorporates best-practice literacy and instructional design to ensure the content and concepts are fully accessible to all students.

Skills and Assessment Book

Pearson Biology 12 New South Wales Skills and Assessment book gives students the edge in preparing for all forms of assessment. Key features include a toolkit, key knowledge summaries, worksheets, practical activities, suggested depth studies and module review questions. It provides guidance, assessment practice and opportunities to develop key skills.



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CHAPTER

04 DNA and polypeptide synthesis

By the end of this chapter, you will be able to describe the differences and similarities of the forms in which DNA exists in eukyarotic and prokaryotic cells. You will also understand the role of nucleic acids (DNA and multiple forms of RNA) as molecules that encode instructions for polypeptide synthesis from amino acids. You will also examine the process of polypeptide synthesis from transcription to translation.

The complexity and importance of proteins will become evident, starting with their construction from polypeptides to the many functions they play in living organisms. You will learn that nearly every function of a living organism depends on proteins.

Content

INQUIRY QUESTION

Why is polypeptide synthesis important?

By the end of this chapter you will be able to:

- construct appropriate representations to model and compare the forms in which DNA exists in eukaryotes and prokaryotes (ACSBL076) ICT
- model the process of polypeptide synthesis, including: (ACSBL079)
  - transcription and translation
  - assessing the importance of mRNA and tRNA in transcription and translation (ACSBL079)
  - analysing the function and importance of polypeptide synthesis (ACSBL080)
  - assessing how genes and environment affect phenotypic expression (ACSBL081) ICT L
- investigate the structure and function of proteins in living things L

4.1 DNA in eukaryotes and prokaryotes

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There are two fundamentally different types of cells—prokaryotic and eukaryotic (Figure 4.1.1). Organisms are classified according to the type of cell that forms their structure. You learnt about the structure of typical prokaryotic and eukaryotic cells in Year 11.

Prokaryotes are usually unicellular, composed of a single prokaryotic cell, and include all species of Bacteria and Archaea. Prokaryotic cells are generally smaller and less complex than eukaryotic cells. The main distinction is that prokaryotic cells have chromosomal **DNA (deoxyribonucleic acid)** but do not hold it in a defined membrane-bound **nucleus** inside the cell.

Eukaryotes are composed of eukaryotic cells and include plants, animals, fungi and protists. They may be unicellular (protists, some fungi) or multicellular (plants, animals and some fungi). Eukaryotic cells contain membrane-bound organelles including the nucleus that contains the DNA inside a double **nuclear membrane**.

In this section, you will learn more about the way DNA is held in each type of cell and the differences and similarities between them.

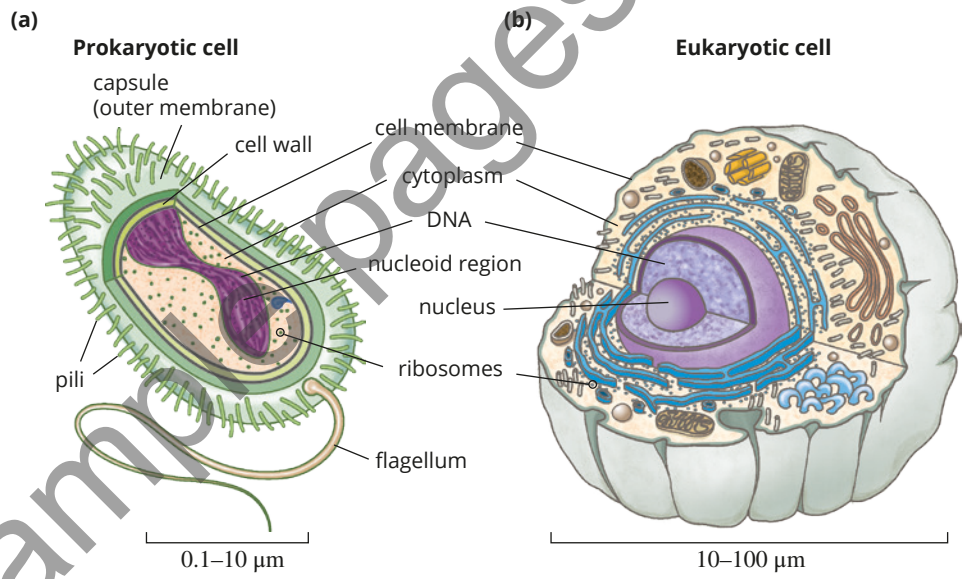

FIGURE 4.1.1 (a) A typical prokaryotic cell compared to (b) a typical eukaryotic cell. Prokaryotic cells and the processes involved in their DNA replication are generally much simpler than eukaryotic cells. Note the different membrane-bound organelles in the eukaryotic cell and the lack of such organelles in the prokaryotic cell.

DNA IN PROKARYOTIC CELLS

Prokaryotic cells are small and lack most membrane-bound organelles, including a distinct nucleus. The term prokaryote actually means 'before nucleus'. Their cytoplasm does contain scattered **ribosomes**, built of two subunits, which are not bound by membranes and are involved in the synthesis of **proteins**. The genetic material of prokaryotic cells is usually one double-stranded, circular DNA **chromosome**, which is contained in an irregularly shaped region called the **nucleoid**. Unlike the nucleus of eukaryotes, the nucleoid is a region of the cytoplasm, not an organelle, and it does not have a nuclear membrane surrounding it.

This circular chromosomal DNA is attached to the inside of the prokaryote's outer cell membrane by a region of the chromosome called the **origin**. Because the DNA is joined in a circle, unlike the linear chromosomes of eukaryotic cells, it does not have the protective end regions called **telomeres** that have a role in the ageing process of a eukaryotic cell.

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Ribosomes in prokaryotic and eukaryotic cells

Ribosomes consist of two subunits joined together (Figure 4.1.2). The subunits in eukaryote ribosomes are different to those in prokaryote ribosomes. Ribosomes are either free in the cytoplasm or bound to rough endoplasmic reticulum (rough ER).

Ribosomes translate mRNA into polypeptides, which then combine to form functional proteins.

The prokaryotic bacteria and archaea have smaller ribosomes, called 70S ribosomes, which are composed of a small 30S subunit and a larger 50S subunit. The S stands for 'svedbergs', a unit used to measure sedimentation speed of molecules in a centrifuge. Eukaryotic ribosomes are larger and consist of a 40S subunit and a larger 60S subunit, which come together to form an 80S particle. The eukaryotic 80S ribosome has a mass of 4200 kd (kilodaltons), compared with 2700 kd for the prokaryotic 70S ribosome. The dalton is a non-SI unit used for mass on the molecular level. One dalton is approximately equal to the mass of one proton or one neutron, which is equivalent to 1 g/mol.

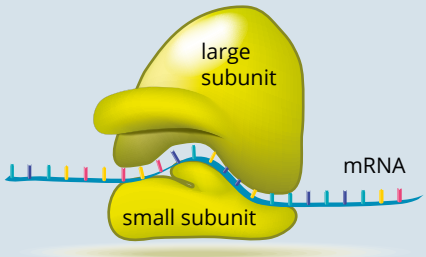

FIGURE 4.1.2 The 80S ribosome in eukaryotic cells is built of a 60S plus a 40S subunit. In prokaryotic cells, the 70S ribosomes are made from 50S plus 30S subunits.

In addition to the chromosomal DNA, many prokaryotic cells also contain small rings of double-stranded DNA called **plasmids**, which can move between cells (Figure 4.1.3). Replication of plasmids is independent of chromosomal replication during binary fission. Plasmids often provide bacteria with genetic advantages, such as antibiotic resistance. From the 1970s, seeing the possibilities in working with a complete small ring of self-replicating DNA, scientists began to use bacterial plasmids in genetic engineering as vectors to clone, transfer and manipulate **genes**. You will learn more about plasmids and their role in genetic technologies in Chapter 9.

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Prokaryotic chromosomes are less condensed than their eukaryotic counterparts (Figure 4.1.4). Unlike eukaryotic chromosomes, the prokaryotic chromosomes do not have features that are easily identifiable when viewed under a light microscope. Usually a stained slide will just show the dark area of the nucleoid.

Most of what we know about the chromosomes of prokaryotes has been obtained from studies of the bacteria, *Escherichia coli*. This species is used as the model organism for much of the research on prokaryotes. It is known that *E. coli* and other bacteria do not have the histone proteins used to condense DNA in eukaryotes. However, some Archaea (the other domain of prokaryotes) do have histones, which is of significance in evolutionary history.

Many prokaryotes also have small hair-like projections called **pili**, which are involved in the transfer of plasmid DNA between organisms by conjugation and can also help generate movement. You learnt about asexual reproduction by conjugation in Chapter 2.

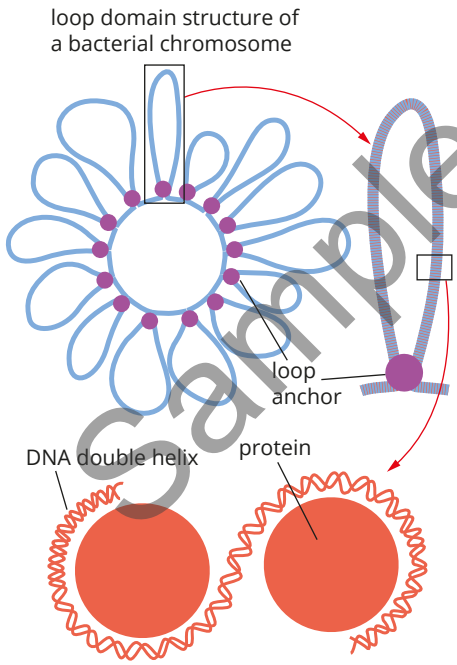

FIGURE 4.1.4 DNA is packaged into loop structures in prokaryotes. The DNA of prokaryotes does not have to be as tightly packaged as in eukaryotes because there is much less genetic material.

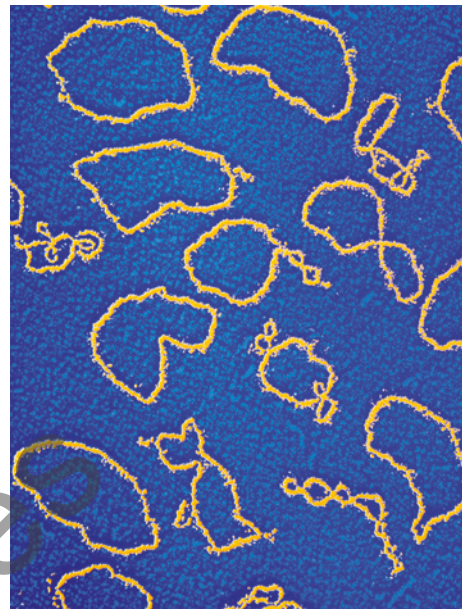

FIGURE 4.1.3 Coloured transmission electron microscope (TEM) of plasmids from the bacteria *E. coli*. Plasmids are independent units, commonly found in prokaryotes, made of small, circular DNA molecules that are much smaller than the single loop of the main chromosome.

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Gene expression in prokaryotes

In prokaryotic cells, **gene expression** consists only of **polypeptide** synthesis and occurs completely within the cytoplasm. Here the two processes of synthesis, **transcription** and **translation**, are closely coupled and occur at almost the same time. The action is between the DNA, **messenger RNA (mRNA)** and ribosomes in the cytoplasm of prokaryotic cells. The two stages involved in synthesis of polypeptide chains will be covered in detail in Section 4.2. Gene expression in prokaryotes is regulated during transcription, the first stage. Unlike eukaryotes, in prokaryotes there is very little processing of RNA, which is the level at which regulation of gene expression takes place for the eukaryotic cells.

DNA IN EUKARYOTIC CELLS

Eukaryotic cells are more complex than prokaryotes (Figure 4.1.1). Eukaryotic cells not only have a cell membrane around the cytoplasm, but also have internal membranes that form specialised membrane-bound compartments within the cell. This is known as cell compartmentalisation. The membrane-bound structures are called organelles. Organelles were covered in detail in Year 11.

REVISION

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Organelles of eukaryotic cells

Organelles are subcellular structures that have a specific function. Because they have a specific function, their presence depends on the needs of the cell. In eukaryotes some organelles, like the nucleus, are membrane-bound and some are not. Prokaryotic cells possess some non-membrane bound organelles, such as ribosomes, flagella and a cell wall, although the structure and composition of these is usually different from that of equivalent eukaryotic organelles.

A eukaryotic cell has a nucleus which contains all of the chromosomal DNA and is wrapped in a double membrane (Figure 4.1.5). The outer nuclear membrane is continuous with the endoplasmic reticulum (ER) that usually holds ribosomes (known as rough endoplasmic reticulum), enabling unobstructed passage for mRNA from the nucleus and transport of the polypeptides after they are synthesised at the ribosomes. Nuclear pores in the membrane permit communication and selective transport between the nucleus and cytoplasm. Within the eukaryotic nucleus there is a smaller organelle called the nucleolus that has a role in forming ribosomes (Figure 4.1.6).

Ribosomes are the tiny organelles that construct polypeptide chains under instruction from the DNA code. In a eukaryotic cell, as in prokaryotes, the ribosome is also built from two subunits but they are both larger than those in prokaryotic cells.

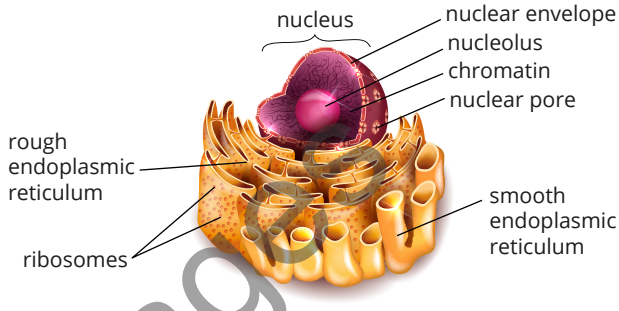

FIGURE 4.1.5 Diagram of the nucleus, nucleolus and rough endoplasmic reticulum (ER) inside a eukaryotic cell. The rough aspect of the ER is actually many ribosomes attached to the ER membrane, which is continuous with the outer nuclear membrane.

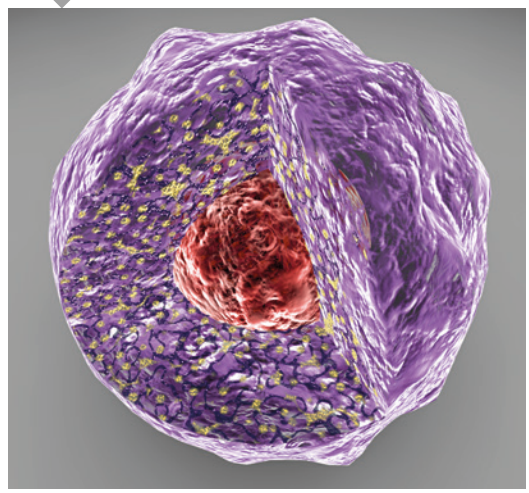

FIGURE 4.1.6 This image shows only the nucleus of a human body cell, with its double membrane dotted with ribosomes on the outside, the dark strands of uncondensed DNA and the nucleolus (red).

Packaging of DNA in eukaryotic cells

In eukaryotic cells, the double-stranded DNA is found in the nucleus where, at times, it is tightly coiled up (condensed) to form the linear structures called chromosomes (Figure 4.1.7). At other stages of the cell cycle, the uncondensed DNA strands form a tightly woven mass. Because eukaryotic cells are more complex than prokaryotes, the eukaryotes have an average of 25 times more DNA than prokaryotes. For it to fit into the nucleus, this amount of DNA has to be much more tightly packaged.

Histones are proteins found in eukaryotic cells that tightly package DNA into structures called nucleosomes. Nucleosomes in turn form supercoils to become the chromosomes.

The processes involved in DNA replication are significantly faster in prokaryotic cells. Some bacterial cells take just 40 minutes to replicate their DNA, while in some animal cells this can take up to 10 hours before cell division.

The structure of DNA and its packaging into nucleosomes, then into linear pairs of homologous chromosomes for eukaryotic cells, has been discussed in Chapter 3. Figure 4.1.7 summarises this information in a diagram.

The tight packaging of chromosomes into the nucleus allows us to view a cell's genetic material under a light microscope.

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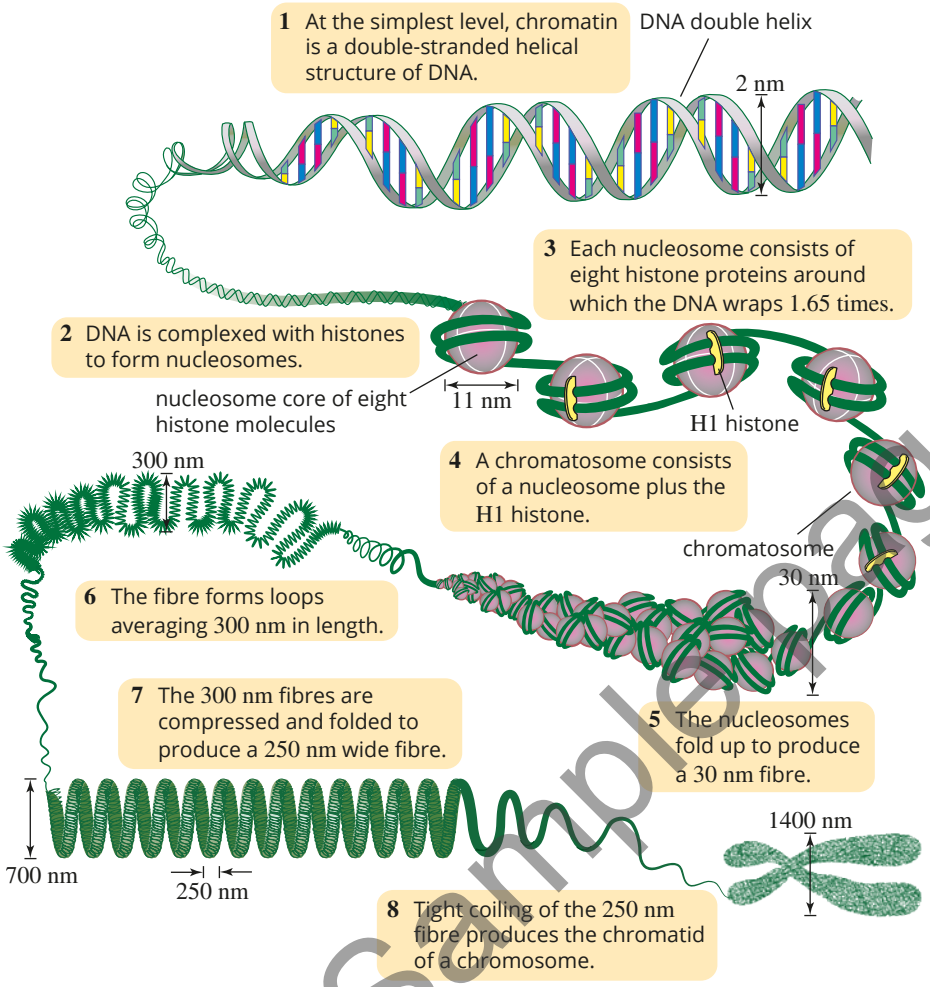

FIGURE 4.1.7 The packaging of DNA in eukaryotic cells. Because eukaryotes have large quantities of DNA to fit into a small space, the DNA needs to be tightly and efficiently packaged. Double-stranded DNA (1) is tightly coiled around histones to form nucleosomes (2 and 3). Nucleosomes and histones together form chromatosomes (4). The nucleosomes fold (5), loop (6) and compress (7) into chromatin. Tight coiling of the chromatin produces the chromatids of a chromosome (8).

A nanometre (nm) is one-billionth of a metre (10^{-9} m). This means there are 1 000 000 (1 million; 10^6) nanometres in a millimetre.

Gene expression in eukaryotes

Gene regulation is tightly controlled in both eukaryotes and prokaryotes. However, as the process of gene expression in eukaryotes is more complex, gene regulation checks occur at a greater number of stages.

In Section 4.2 you will learn that gene expression in eukaryotes comprises the processes of transcription, **RNA processing** and translation. Gene expression can be regulated at any of these stages. In eukaryotic cells, transcription and RNA processing occur within the nucleus, and translation occurs in the cytoplasm.

Gene expression in prokaryotes consists only of transcription and translation, whereas in eukaryotes, it involves transcription, RNA processing and translation.

COMPARISON OF PROKARYOTIC AND EUKARYOTIC DNA

Both types of cells contain double helix DNA as their inherited genetic material, making DNA universal in structure and function to all living things on our planet. An essential difference between prokaryotes and eukaryotes is the way the DNA molecule is packaged inside their cells.

Similarities

Although there are many differences between the gene structures and DNA packaging of prokaryotes and eukaryotes, shared evolutionary history means that there are also many fundamental similarities.

- Both prokaryotes and eukaryotes have double-stranded DNA, twisted into a double helix and built from the same type of nucleotides with the same nitrogenous base molecules—adenine (A), thymine (T), cytosine (C) and guanine (G).
- Both have mRNA, which acts as an intermediate code to building proteins, with the base uracil (U) replacing thymine (T) in the mRNA.
- Because the genetic information of prokaryotes and eukaryotes is composed of the same code, the way mRNA is translated into **amino acids** and proteins is also much the same. The universality of the DNA and mRNA coding has made the use of prokaryotic plasmids possible as vectors for genetic engineering across all species. Examples of the use of plasmid vectors can be found in Chapter 9. Section 4.2 will cover polypeptide synthesis in more detail.

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Differences

Prokaryotic and eukaryotic genes are structurally different in several ways. These differences affect the way in which genetic information is transcribed, translated and expressed. Table 4.1.1 summarises the major differences between the structure of the genetic material of prokaryotes and eukaryotes.

TABLE 4.1.1 Summary of the major differences between the structure of the genetic material of prokaryotes and eukaryotes.

Prokaryote	Eukaryote
Chromosomal DNA is in a region of cytoplasm called the nucleoid, lacking a membrane. There is one chromosome per cell.	Chromosomal DNA is in the nucleus, which is separated from the cytoplasm by a double-layered membrane. There are multiple chromosomes per cell of a diploid ($2n$) or haploid (n) number.
A circular chromosome without ends (no telomeres).	Linear thread-like chromosomes with ends (telomeres).
Contains plasmids—small, circular DNA.	Contains no plasmids but there are other sources of DNA apart from chromosomes—mitochondrial DNA and chloroplast DNA.
There is much less DNA than in eukaryotes (thousands to millions of bases, depending on species).	There is much more DNA than in prokaryotes (millions to billions of bases, depending on species).
There are fewer genes than in eukaryotes (thousands).	There are more genes than in prokaryotes (tens of thousands).
There is less non-coding DNA (introns) than in eukaryotes (greater number of genes per number of bases).	There is more non-coding DNA (introns) than in prokaryotes (fewer genes per number of bases).
DNA is in a region called the nucleoid but is not packaged into an organelle (less DNA to fit into the cell).	DNA is tightly packaged—coiled around histones forming nucleosomes, which are condensed into chromatin and packed as chromosomes into the nucleus (a lot of DNA to fit into a small space).
Genes cluster into functional groups, known as operon regions (e.g. genes that code for enzymes in the same biochemical pathway are next to each other on the chromosome, so all the genes for the pathway can be transcribed and expressed at once).	Genes that code for functionally similar proteins can be physically far apart or located on different chromosomes. Eukaryotes have mechanisms to express these genes at the same time.

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To code or not to code

The role of DNA in both prokaryote and eukaryote cells is to carry the inherited code (genes) for production of polypeptides that combine into the many essential protein molecules required by a cell. Prokaryotes need to carry all their coding genes in one relatively short chromosome, so there is not much space for non-coding sequences of DNA. In eukaryotes, the amount of non-coding lengths of DNA is about 98% compared to prokaryotes where it is only 12%. The coding segments of DNA are known as exons and the non-coding regions as introns (Figure 4.1.8).

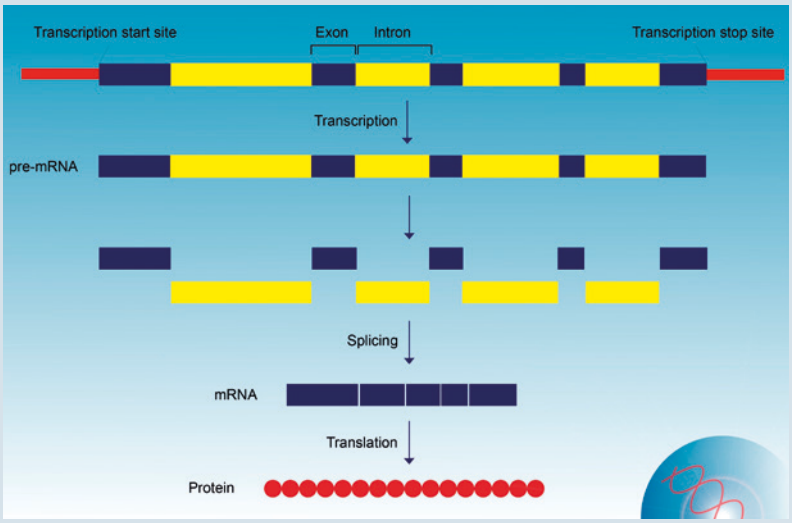


FIGURE 4.1.8 Gene codes for producing proteins are carried in the exon regions of a chromosome. Introns are the non-coding regions and there are far less of these in prokaryote cells. Some of the processes depicted in this diagram are explained in Section 4.2.

BIOLOGY IN ACTION CCT ICT

The first genome

The first free-living organism to have its **genome** sequenced was a bacterium, *Haemophilus influenzae*. The work was completed in 1995 at TIGR (The Institute for Genomic Research, now called J. Craig Venter Institute) in the USA. The prokaryotic genome consists of about 1.8 million base pairs in a single circular chromosome. It was sequenced in 13 months at a cost of 50 cents (US) per base unit. At the time this was a record for speed and cost, now it is surpassed by vastly improved methods.

The rapid advancement of microchips and processors capable of managing and storing the vast amount of data, even from this relatively short, simple genome, was the key to completing the groundbreaking project. TIGR had developed new sequencing techniques and computational methods, as well as improved data storage and hardware. Software called the TIGR Assembler was used to assemble approximately 24 000 DNA fragments into the whole genome. At the time, the software required approximately 30 hours of processing time with half a gigabyte of RAM, an indication of the complex computation involved.

FIGURE 4.1.9 Zebrafish (*Danio rerio*) are freshwater aquarium fish that are commonly used as model organisms. Their eukaryotic genome was sequenced in 2013.

In 2003, the Human Genome Project was completed. This ushered in a new era, known as Next Generation Sequencing (NGS). Bioinformatics was born, a field where computer science and biology operate together.

Now, thanks to improved sequencing machines with increased accuracy and speed, the genomes of many eukaryotic species have also been sequenced. By 2013, the zebrafish (*Danio rerio*) (Figure 4.1.9) genome with 1.5 billion bases and 25 chromosomes, had been sequenced. More numerous and increasingly more complex genomes are being sequenced every month. Automation and digital technology with improved capacity are rapidly moving the field of genomics research forward, lowering costs and providing faster analysis.

The new era of bioinformatics has been likened to the establishment of the periodic table in the 19th century as a tool for chemists. Modern geneticists can now construct readily accessible databases of genes and DNA sequences (Figure 4.1.10).

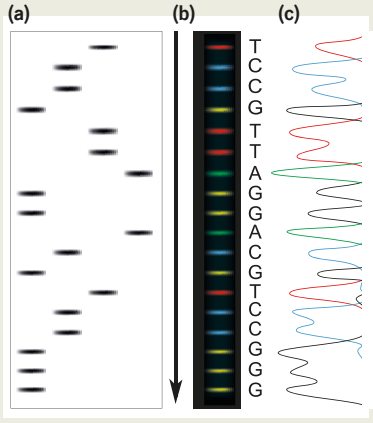


FIGURE 4.1.10 Illustration of the basic data from DNA sequencing. (a) capillary electrophoresis to separate nucleotide fragments, (b) colour fluorescence in a laser beam and (c) chromatogram generated by computer software.

4.1 Review

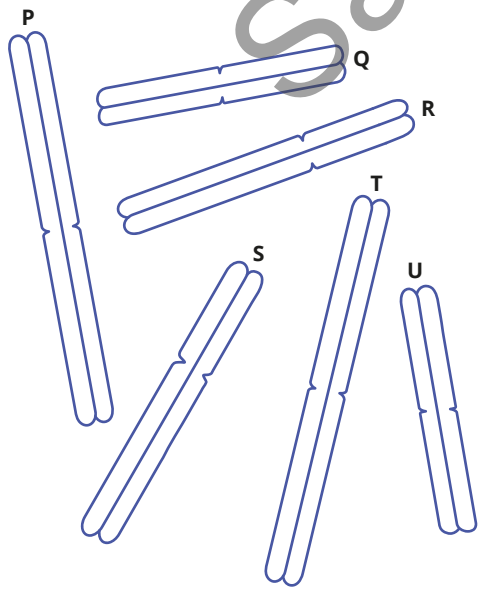
SUMMARY

- Organisms with prokaryotic cells are called prokaryotes. They are classified into two domains—Bacteria and Archaea.
- Prokaryotic cells are small, with simple structure and lack membrane-bound organelles. They have scattered ribosomes and a nucleoid region containing DNA.
- Prokaryotic DNA is carried as one chromosome that is a continuous loop of double-stranded DNA in the nucleoid region plus small rings of DNA called plasmids that operate independently in the cytoplasm.
- The single loop of chromosomal DNA in prokaryotic cells is formed mainly of coding sequences (exons).
- Organisms with eukaryotic cells are called eukaryotes. They are classified into the domain Eukarya, which is divided into four kingdoms: Protista, Fungi, Plantae and Animalia.
- Eukaryotic cells have a complex structure with a membrane-bound nucleus, many organelles in the cell cytoplasm, and DNA mainly in linear chromosomes in the nucleus.
- There are many differences in the structure of the genetic information of prokaryotes and eukaryotes (e.g. prokaryotes have much less DNA but more genes per number of bases than eukaryotes). Because eukaryotes have much more DNA, they package it more tightly. These structural differences affect the way genetic information is transcribed, translated and expressed into proteins.
- The ribosomes of both cell types are formed of two subunits with eukaryotes having the larger 80S ribosomes and prokaryotes having 70S ribosomes.
- Although there are many differences, the basic genetic structures of prokaryotes and eukaryotes share many similarities. For example, the same code of nitrogenous bases (adenine [A], thymine [T], cytosine [C], guanine [G] and uracil [U]) translates into amino acids, polypeptides and proteins in much the same way for both prokaryotes and eukaryotes.

KEY QUESTIONS

1 Use labelled diagrams to illustrate the main differences between prokaryotic and eukaryotic cells. Identify an example of each.

2 The figure below shows six chromosomes belonging to three homologous pairs in a eukaryotic cell.



a Identify the three pairs of homologous chromosomes.

b Describe which features you used to match the chromosomes.

3 A human cell has approximately 25 000 genes and *E. coli* has approximately 4000 genes. Explain why the number of genes is not an indicator of the size of the genome.

4 Summarise five differences between prokaryotic and eukaryotic gene structure.

5 Examine some of the main differences in gene regulation between prokaryotic and eukaryotic cells.

6 Explain why the chromosomes of eukaryotic cells can be seen under a light microscope but those of prokaryotes cannot.

4.2 Polypeptide synthesis

The **genetic code** represents the inherited genetic information stored in DNA as triplet bases within sections called genes. This information is transcribed to RNA, then used to synthesise the amino acid sequences that form polypeptides through a process called gene expression. Chains of polypeptides combine to form proteins (Figure 4.2.1). Proteins are biological molecules that carry out most of the functions that are essential to life. Antibodies for immune response, collagen for tissue repair, enzymes that catalyse biochemical reactions, hormones for cellular signalling and haemoglobin for transport are all types of proteins.

In this section, you will learn about the roles of DNA and RNA in polypeptide synthesis, the different steps of gene expression and that functional proteins are formed from polypeptides. You will begin to appreciate the significance of protein molecules in a living organism, an area that will be further developed in Section 4.3. You may find it useful to revise the structure of nucleic acids from Chapter 3 before continuing with this section. The influence of the environment on phenotypic expression will also be explored.

Polypeptide means 'many peptide bonds'. A protein is formed by one or more polypeptides arranged in a biologically functional way.

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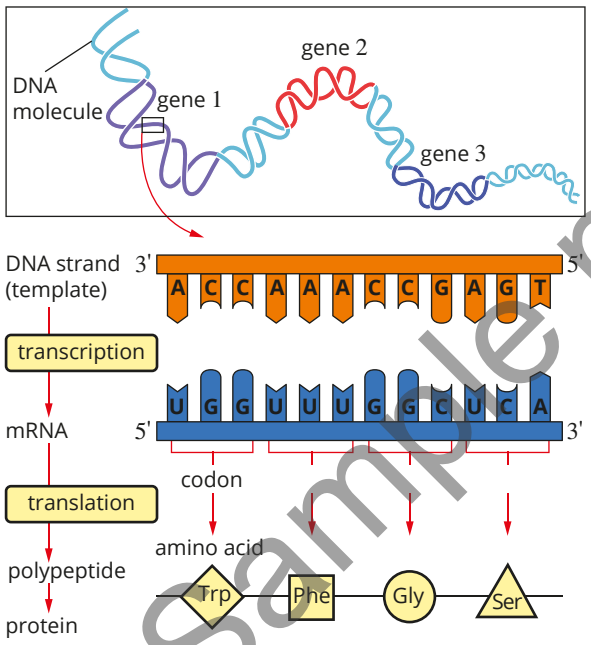

FIGURE 4.2.1 A model diagram for polypeptide and protein synthesis

THE GENETIC CODE

The genetic code is a set of rules that defines how the information in DNA and RNA is translated into polypeptides, which join to form proteins. The genetic code is universal—the rules are the same for all organisms on Earth.

As you learnt in Chapter 3, the information in DNA and RNA is stored as a three-letter code of nucleotides. RNA works in partnership with DNA to synthesise polypeptides as required by a eukaryotic cell. In this section you will learn about the roles of these nucleic acids in the synthesis of polypeptides.

Both DNA and RNA are made up of nitrogenous bases and a sugar–phosphate backbone. RNA is single-stranded and relatively short. DNA has a double-stranded helix (spiral) structure with complementary pairing of its nitrogenous bases holding the double strands together like rungs on a ladder.

RNA contains a pyrimidine base called uracil (U) in place of thymine (T) in DNA. Adenine (A) in DNA is transcribed into uracil (U) in mRNA.

Each DNA triplet or codon codes for one amino acid and may also provide specific instructions, such as 'start translation' and 'stop translation'.

Role of DNA in polypeptide synthesis

DNA provides the instructions, which are translated by RNA into polypeptides and proteins. DNA stores and transmits hereditary information as a sequence of nucleotides. The order of the nucleotides in DNA determines which polypeptides are synthesised. Groups of three nucleotides are called **triplets**. When a DNA triplet is transcribed into mature mRNA, the triplet is then called a **codon** (Figure 4.2.2). For polypeptide synthesis to occur, DNA and RNA must function together.

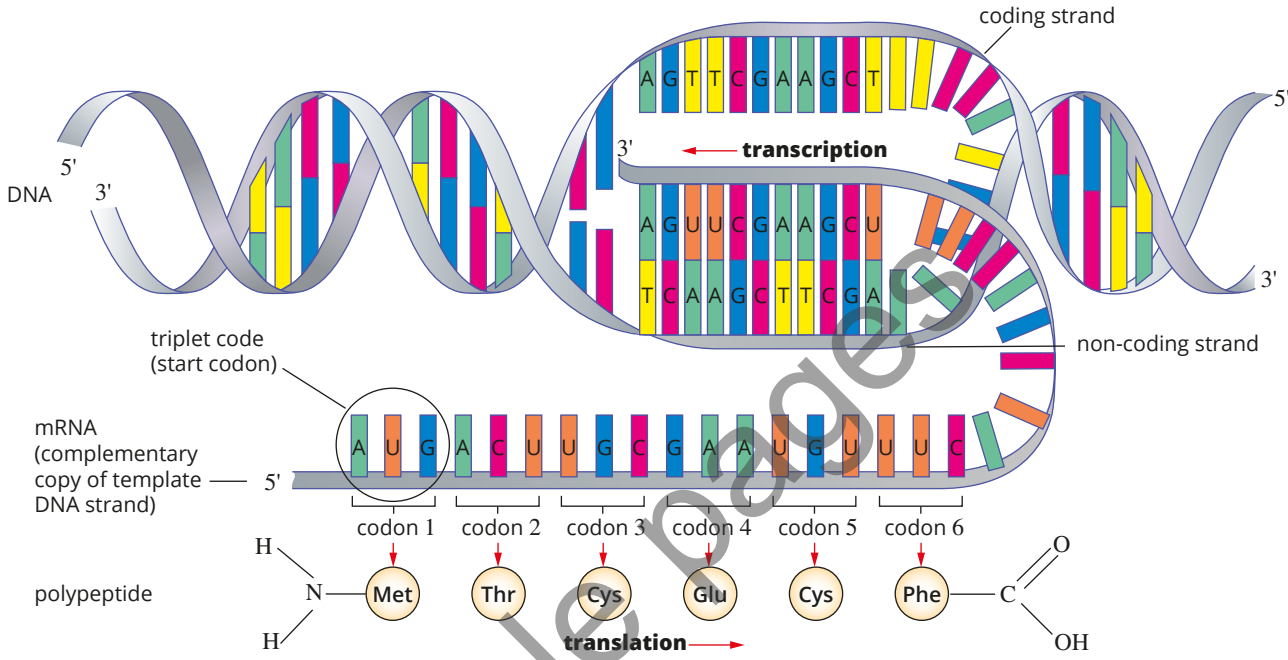

FIGURE 4.2.2 The DNA triplets are transcribed into messenger RNA (mRNA). mRNA is read as codons. Each codon codes for a particular amino acid with the exception of stop codons that end protein synthesis.

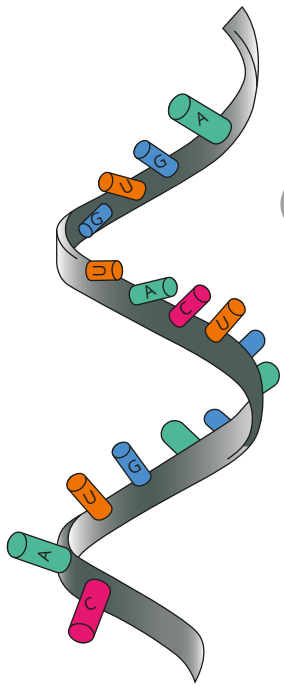

FIGURE 4.2.3 Messenger RNA (mRNA) carries a copy of the DNA's nucleotide sequence to be translated into proteins.

Role of RNA in polypeptide synthesis

RNA plays an important role in expressing the information contained in the DNA sequence to synthesise polypeptides. There are three main forms of RNA and each has a different role in polypeptide synthesis:

- Messenger RNA (mRNA) is formed in the nucleus by the process of transcription. The mRNA carries a complementary copy of the nucleotide sequence of DNA that specifies the amino acid sequence for a particular polypeptide (Figure 4.2.3). During transcription, the primary transcript (pre-mRNA) is first formed by the enzyme **RNA polymerase**. Pre-mRNA is then processed (post-transcriptional modification) to form mature mRNA, which is a single-stranded complementary copy of the coding DNA (gene or **exon**) (Figure 4.2.4). The mature mRNA travels from the nucleus to the cytosol where it binds to ribosomes ready for translation.
- **Ribosomal RNA (rRNA)** is synthesised in the nucleolus of the cell nucleus and is based on the nucleotide sequence of the DNA. Together with proteins, rRNA forms a small organelle called a ribosome. Ribosomes are the sites where the information in the mRNA is translated into a chain of amino acids (Figure 4.2.5).
- **Transfer RNA (tRNA)** molecules transfer amino acids from the cytoplasm to the ribosomes, where they are joined to form a polypeptide chain based on the sequence of nucleotides in the mRNA (Figures 4.2.5 and 4.2.6).

There are 61 different tRNA molecules, each of which combines with only one particular amino acid at one end of its molecule. (There are 64 codons that each represent an amino acid, three of which are **stop codons**. There are no tRNA molecules that recognise these stop codons and so translation is terminated.) There are three places for tRNA to bind to the ribosome: the exit site (E), the peptidyl site (P) and the aminoacyl (A) as shown in Figure 4.2.6b. At the other end of the tRNA molecule, there is a sequence of nucleotides known as the **anticodon**. The anticodon recognises a particular sequence of nucleotides in the mRNA. This enables an amino acid to be positioned in the correct place on a polypeptide chain.

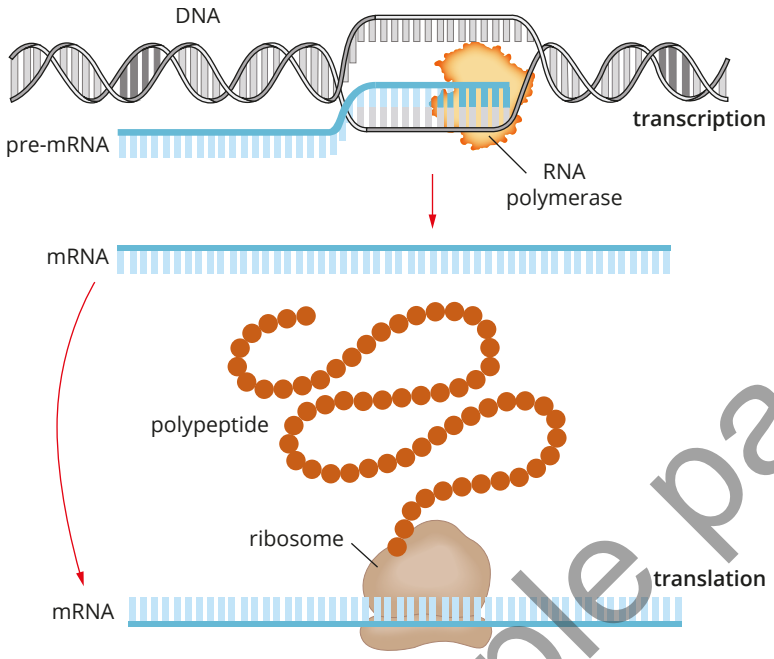

FIGURE 4.2.4 During transcription, RNA polymerase makes a complementary copy of the DNA, which first becomes pre-mRNA and then mature mRNA. The mRNA is then transported to the ribosomes where it is translated into a chain of amino acids, making a polypeptide.

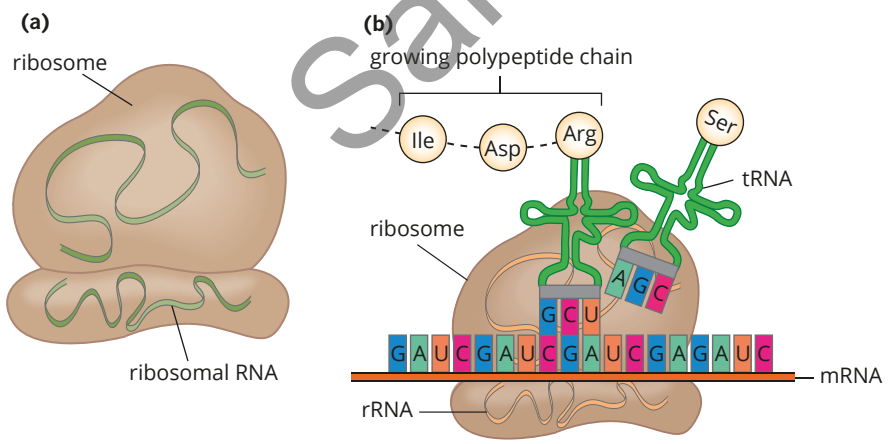

FIGURE 4.2.5 (a) Ribosomal RNA (rRNA), together with two protein subunits, forms ribosomes that are the site of translation of the mRNA into polypeptides. (b) The three different types of RNA (mRNA, rRNA and tRNA) work together to use the information contained in a DNA gene to synthesise a polypeptide.

The primary transcript is the first RNA product synthesised during DNA transcription. The primary transcript undergoes processing to form mature, functional RNA products (e.g. mRNA, rRNA and tRNA). The primary transcript of mRNA is known as pre-mRNA.

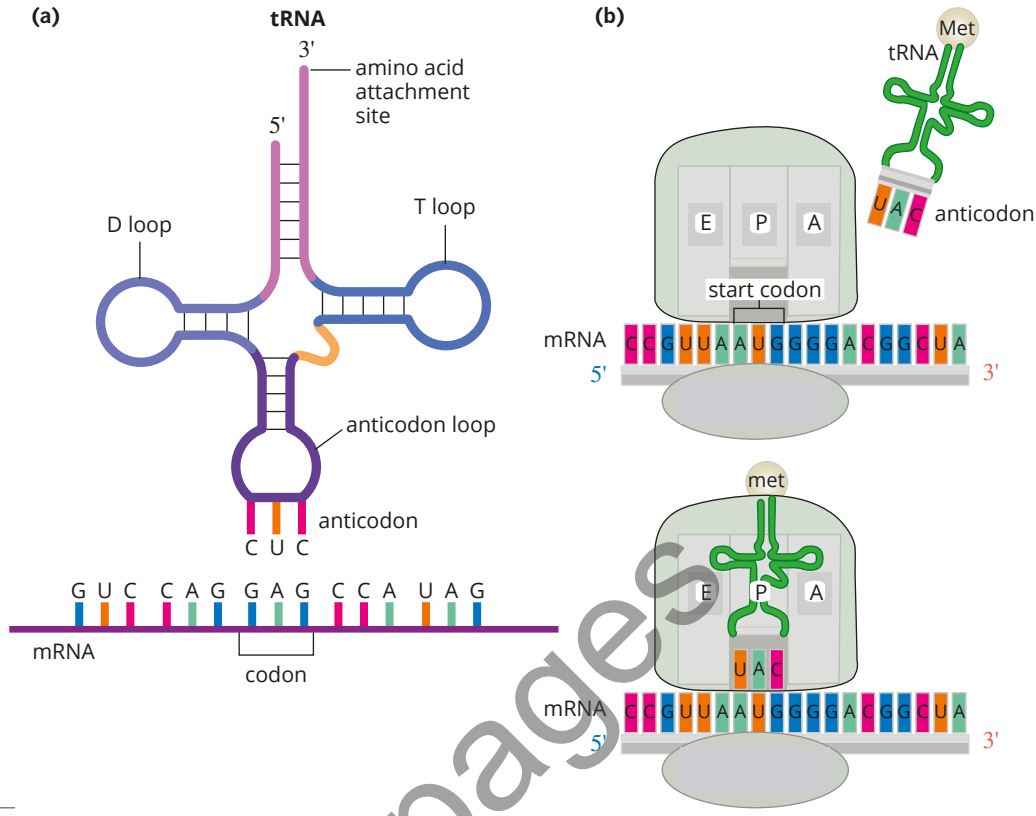

FIGURE 4.2.6 (a) Transfer RNA (tRNA) carries amino acids to the appropriate positions on the mRNA by matching its anticodon sequence to the complementary sequence in the mRNA. The amino acids transferred by tRNA build the polypeptide chain during translation. (b) In this figure, a tRNA molecule carrying the amino acid methionine (Met) recognises the codon on mRNA that is complementary to its anticodon sequence and transfers the amino acid in the correct position on the mRNA. Met is a start codon and the first amino acid to be incorporated into the polypeptide chain.

The genetic code for determining amino acid sequences works in sets of three bases (nucleotides): on DNA, the set of three is called a triplet; on mRNA, it is called a codon; on tRNA, it is an anticodon. For example, the coding for lysine is: DNA triplet TTC, mRNA codon AAG, tRNA anticodon UUC (remembering that RNA has U instead of T).

Degeneracy of the genetic code

The genetic code is said to be degenerate because more than one RNA codon can code for the same amino acid (Figure 4.2.7). Differences in codons encoding the same amino acid usually occur at the second or third base. As the genetic code uses four nucleotides and three nucleotides code for an amino acid, the combinations of these nucleotides make a total of 64 possible codons ($4^3 = 64$), to code for the total 20 amino acids (Figure 4.2.7). The degeneracy of the code acts as a buffer against mutations in that a single change in one base may not necessarily lead to a change in the amino acid produced. Therefore, it may not necessarily change the structure of the protein produced.

Second base of codon

First base of codon	U		C		A		G		Third base of codon
U	UUU	phenylalanine (Phe)	UCU	serine (Ser)	UAU	tyrosine (Tyr)	UGU	cysteine (Cys)	U
	UUC		UCC		UAC		UGC		C
	UUA	leucine (Leu)	UCA		UAA	STOP	UGA	STOP	A
	UUG		UCG		UAG		UGG	tryptophan (Trp)	G
C	CUU	leucine (Leu)	CCU	proline (Pro)	CAU	histidine (His)	CGU	arginine (Arg)	U
	CUC		CCC		CAC		CGC		C
	CUA		CCA		CAA	glutamine (Gln)	CGA		A
	CUG		CCG		CAG		CGG		G
A	AUU	isoleucine (Ile)	ACU	threonine (Thr)	AAU	asparagine (Asn)	AGU	serine (Ser)	U
	AUC		ACC		AAC		AGC		C
	AUA		ACA		AAA	lysine (Lys)	AGA	arginine (Arg)	A
	AUG	methionine (Met) START	ACG		AAG		AGG		G
G	GUU	valine (Val)	GCU	alanine (Ala)	GAU	aspartic acid (Asp)	GGU	glycine (Gly)	U
	GUC		GCC		GAC		GGC		C
	GUA		GCA		GAA	glutamic acid (Glu)	GGA		A
	GUG		GCG		GAG		GGG		G

FIGURE 4.2.7 The genetic code for the 20 amino acids and stop codons. Remember that these are mRNA codons (not DNA triplets), a fact that should be apparent as soon as U for uracil is seen in the list of bases. The start codon methionine (Met) is highlighted yellow and the stop codons are highighted grey.

Worked example 4.2.1

UNLOCKING THE GENETIC CODE

Complete the table by entering the mRNA codons and the complementary DNA triplet bases that code for each amino acid listed.

Amino acid	mRNA codons	Complementary DNA triplets
Ala alanine		
Lys lysine		

Thinking	Working
Determine how to read the genetic code in Figure 4.2.7. A set of three bases in mRNA is a codon for one amino acid.	The codon is read first from the left column, then the top row, then the right column (e.g. alanine (Ala) is coded by G then C then U or C or A or G).
Remember that U replaces T in mRNA. The complementary DNA base for U is A.	One of the mRNA codons for alanine is GCU (not GCT) and the complementary DNA will be CGA.
Remember there are two to four repeat codons for each amino acid (64 codons and 20 amino acids). This is called degeneracy.	Alanine has four codons: GCU, GCC, GCA, GCG Lysine has two codons: AAA, AAG Enter the mRNA codons into the table: **Amino acid** / **mRNA codons** / **Complementary DNA triplets** Ala alanine / GCU, GCC, GCA, GCG / Lys lysine / AAA, AAG /
Identify the complementary triplet codes (base sequence) of the DNA that these mRNA codons were transcribed from.	Alanine codons are: GCU, GCC, GCA, GCG Complementary DNA triplets are: CGA, CGG, CGT, CGC Lysine codons are: AAA, AAG Complementary DNA triplets are: TTT, TTC Enter the complementary DNA triplets into the table: **Amino acid** / **mRNA codons** / **Complementary DNA triplets** Ala alanine / GCU, GCC, GCA, GCG / CGA, CGG, CGT, CGC Lys lysine / AAA, AAG / TTT, TTC

Worked example: Try yourself 4.2.1

Complete the table by entering the mRNA codons and the complementary DNA triplet bases that code for each amino acid listed.

Amino acid	mRNA codons	Complementary DNA triplets
Arg arginine		
Asn asparagine		
Asp aspartic acid		
Cys cysteine		
His histidine		
Leu leucine		
Tyr tyrosine		
Val valine		

THE STRUCTURE OF GENES

While the genetic code is universal, the structure of genes and chromosomes differs between prokaryotes and eukaryotes (see Section 4.1). Prokaryotes contain fewer introns than eukaryotes, which simplifies the process of polypeptide synthesis in prokaryotic cells.

Eukaryotic genes all have structural features in common, including:

- stop and start triplet sequences—regions where encoding DNA begins and ends for a specific gene
- **promoter**—sections of a gene that are found on the DNA before the start triplet (**start codon**), at the 5' end of the site where transcription will begin. The promoter is the location where the RNA polymerase (the enzyme that initiates transcription) attaches to the gene. In many eukaryotic genes, the promoter region is coded for by the sequence of bases TATAAA, which is sometimes called the TATA box
- exons—DNA regions that are the coding segments
- **introns** (or spacer DNA)—DNA regions that are non-coding segments (Figure 4.2.8).

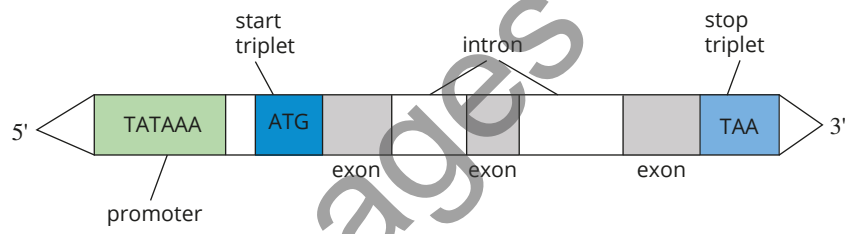

FIGURE 4.2.8 Eukaryotic genes have promoter regions, start and stop triplets, non-coding introns and coding exons.

The information on the coding strand of DNA is one side of the double helix. It is this information that is transcribed into mRNA ready for translation into proteins.

+ ADDITIONAL

Start and stop instructions

A start codon indicates where the first stage of gene expression will begin. AUG (the amino acid methionine) is the most common start codon in mRNA. In DNA, the start triplet coding for AUG is written as ATG (Figure 4.2.8). This is because when writing a DNA sequence, the scientific standard is to show only the coding (non-template) strand of DNA and to write it in the 5' to 3' direction. The triplet that is complementary to AUG (TAC) is found on the non-coding (template) strand of DNA, which is read by RNA polymerase when building mRNA (see Figure 4.2.2). When transcribed into mRNA the DNA triplet ATG will become the start codon AUG. This codon initiates translation and codes for the amino acid methionine. Most functional proteins start with AUG, but there are some rare exceptions to this. For example, a protein in the fungus, *Candida albicans*, uses GUG as a start codon.

A stop triplet indicates where transcription will end. The stop triplet does not code for an amino acid. When the stop triplets are transcribed into mRNA they become the codons UAA, UAG and UGA which are not recognised by tRNA.

Figure 4.2.7 provides the full set of genetic codes for the 20 amino acids and start/stop codons. Remember that each amino acid corresponds to three bases of mRNA (a codon), not DNA triplets.

Introns and exons

In eukaryotes, not all DNA regions are translated.

- Exons are DNA regions that are usually 'expressed' as proteins or RNA. Exons come together to make up mRNA, which is then translated into proteins.
- Introns are non-coding, or intervening, regions of DNA. Introns are spliced out of the mRNA during RNA processing.

In models of nucleic acids, the nucleotides are simply referred to as bases and are identified by the base letter, A, T, C, G or U. This is because the sugar and phosphate units in all nucleotides are identical. The nitrogenous base is the unit that changes.

GENE EXPRESSION

Gene expression is the process by which the information stored in a gene synthesises a functional gene product (protein or RNA). This process is highly regulated so that proteins or RNA molecules are only produced if they are required by a cell. Multicellular organisms in particular have specialised cells that require a specific set of proteins. For example, in humans, the cells in connective tissue and bone require the protein fibrillin to form elastic fibres, and skin cells require the enzyme tyrosinase to produce melanin and other pigments. The ability to regulate gene expression conserves energy and materials (e.g. nucleotides and amino acids) in the cell.

Gene expression leading to polypeptide synthesis in eukaryotic cells occurs in three stages:

- transcription
- RNA processing
- translation.

In eukaryotic cells, transcription and RNA processing occur within the nucleus and translation occurs in the cytoplasm (Figure 4.2.9).

The genetic code is always a three-letter code, read as groups of three bases. For example, AGU is an mRNA codon for the amino acid called serine. This codon was transcribed from the complementary DNA triplet of TCA. And the matching tRNA anticodon will be UCA.

Messenger RNA (mRNA) is produced during transcription and then translated to produce an amino acid chain (polypeptide).

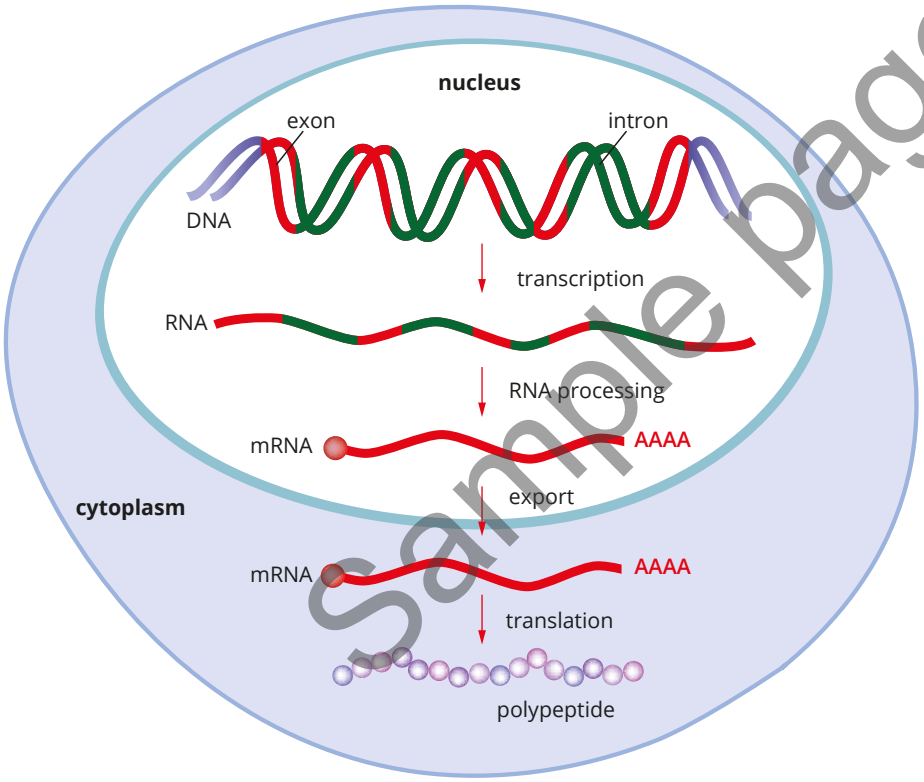

FIGURE 4.2.9 Transcription creates a primary transcript from DNA. The introns are then spliced (cut out) during RNA processing to create a mature strand of mRNA. The mRNA exits the nucleus via a nuclear pore. A ribosome translates the mRNA into a polypeptide chain during translation.

Transcription

Producing single-stranded mRNA from DNA is called transcription and occurs within the nucleus of eukaryotic cells. The DNA segment that undergoes transcription is known as the transcription unit.

Transcription occurs in three steps:

1. initiation
2. elongation
3. termination (Figure 4.2.10).

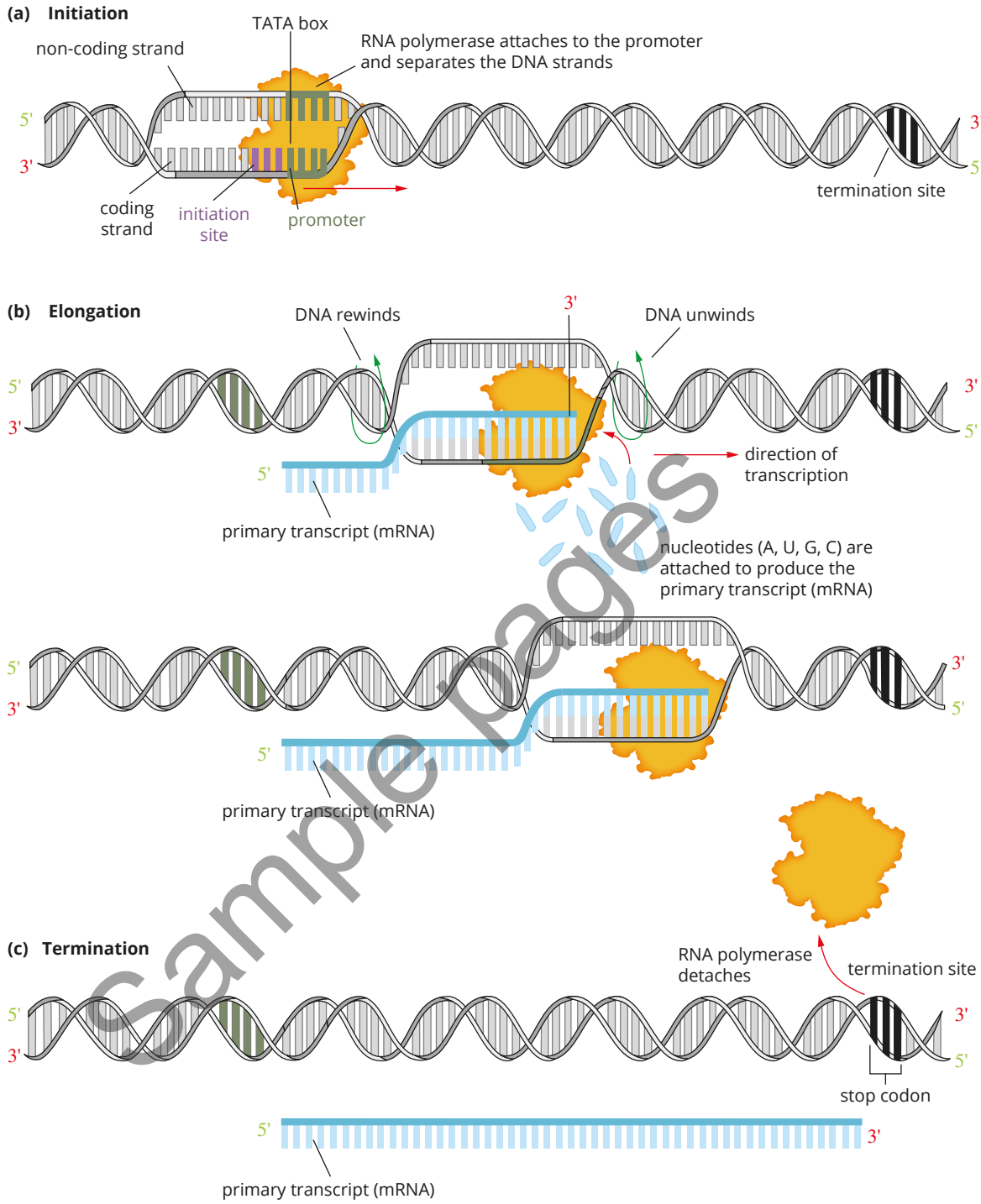

FIGURE 4.2.10 Transcription occurs in three stages. (a) Initiation of transcription, when RNA polymerase attaches to the promoter region of the DNA and unzips the two DNA strands by breaking their hydrogen bonds, exposing the nitrogenous bases (A, T, G, C). (b) During elongation, the RNA polymerase moves along the coding DNA strand and produces a strand of mRNA by attaching complementary nucleotides (A, U, G, C). The mRNA strand is known as the primary transcript (or pre-mRNA) at this stage. (c) Transcription is terminated when the RNA polymerase reaches the stop triplet code (stop codon) at the termination site. The RNA polymerase then detaches and the two DNA strands come together.

Many RNA polymerase molecules may attach to the gene being transcribed, producing many of the same mRNA molecules. The strand of DNA that is transcribed to the mRNA is known as the **non-coding strand** (or template strand) and the other complementary strand is known as the **coding strand** (or non-template strand). The mRNA carries the same base sequence as the coding strand, (except it contains uracil in place of thymine) because it has been copied from the non-coding strand into complementary bases.

RNA processing

After transcription, the primary RNA transcript is processed before being translated. RNA processing forms mature mRNA from pre-mRNA (the primary transcript of mRNA), after removing non-coding sequences (introns) so that only the coding sequences (exons) are carried to the ribosome for translation (Figure 4.2.11). RNA processing is present in both eukaryotic and prokaryotic cells but is much more complex in eukaryotes, as prokaryotes carry fewer introns that need to be spliced out after transcription. The RNA processing stage of gene expression in eukaryotes includes:

- the addition of a **5' cap**
- the addition of a **poly(A) tail** (The 5' cap and poly(A) tail make the mRNA more stable and prevent it from degrading.)
- **splicing** (removal) of the introns (mRNA maturation).

For prokaryotes, without a nuclear membrane, the transcription and translation processes are closely coupled in the cytoplasm, with the mRNA moving off the DNA and becoming attached to ribosomes even before transcription is complete.

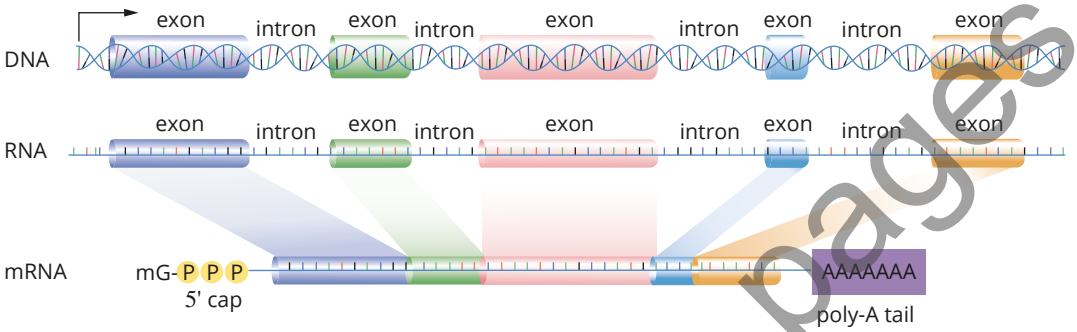

FIGURE 4.2.11 During RNA processing, the introns (non-protein-coding regions) are spliced from the primary pre-mRNA transcript, resulting in mature messenger RNA, which consists of only exons (protein-coding regions).

Translation

Translation is the process in which the codons on mRNA are translated into a sequence of amino acids resulting in a polypeptide. This process occurs on ribosomes in the cytoplasm. Ribosomes bind to an mRNA molecule and act as docking stations for the tRNAs to deposit their specific amino acids. A part of the tRNA, called an anticodon, recognises and binds to the codon on the mRNA by complementary base pairing. Each tRNA carries a specific amino acid related to the codon to which it binds.

Like transcription, translation also occurs in a series of three steps:

1 initiation
2 elongation
3 termination (Figure 4.2.12).

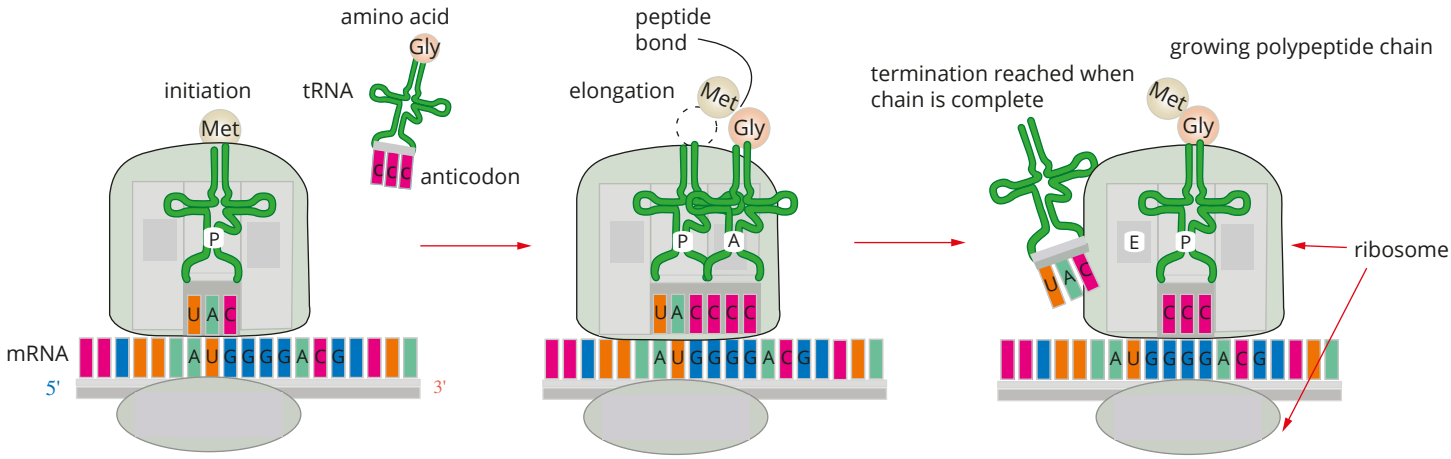

FIGURE 4.2.12 The process of translation on a ribosome. The ribosome moves along the mRNA one codon at a time and tRNA molecules bring their specific amino acids to their complementary mRNA codon. The amino acids join together by peptide bonds to form a polypeptide chain.

Many ribosomes can translate the same, single strand of mRNA, enabling many polypeptide chains to be produced at the same time (Figure 4.2.13). Once the polypeptides are fully functional, they either remain in the cell for use, or are exported from the cell by vesicles (via a process called exocytosis) for use elsewhere in the organism.

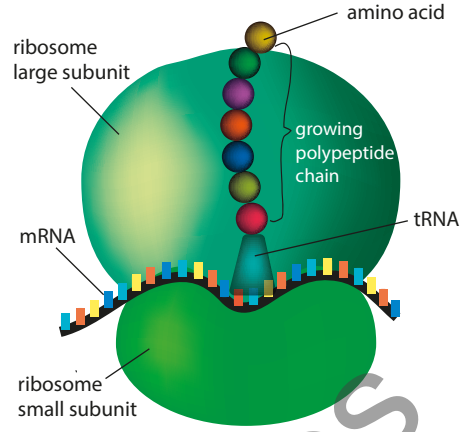

FIGURE 4.2.13 Eukaryotic ribosome translating mRNA into a polypeptide

Protein synthesis in prokaryotes

Prokaryotes do not have membrane-bound organelles, so all cellular processes occur within the cytosol. This allows transcription and translation to be a continuous process rather than two separate stages. Ribosomes can attach to the mRNA while it is being transcribed, so translation can occur at the same time. Prokaryotes mostly contain exons, so splicing rarely occurs before translation.

There are many differences between protein synthesis in prokaryotic and eukaryotic cells (Figure 4.2.14). Table 4.2.1 summarises the major differences, many of which have been used to develop drugs that target protein synthesis in prokaryotes only. For example, some antibiotics disrupt or inhibit the production of proteins in disease-causing bacteria.

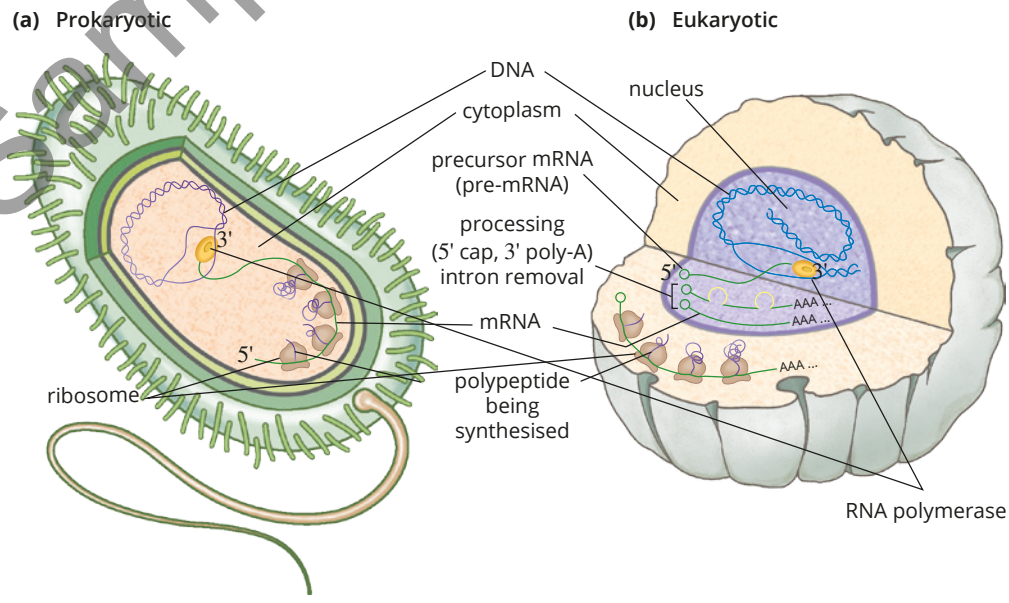

FIGURE 4.2.14 Comparison of protein synthesis in (a) prokaryotic and (b) eukaryotic cells. The structural differences between the cell types means that there are many differences in the way proteins are synthesised.

TABLE 4.2.1 A summary of the differences between prokaryotic and eukaryotic protein synthesis

Prokaryotic protein synthesis	Eukaryotic protein synthesis
30S and 50S ribosomal subunits (forms 70S ribosome)	40S and 60S ribosomal subunits (forms 80S ribosome)
Each mRNA may contain the coding sequences of several genes; this is known as polycistronic.	Each mRNA contains the coding sequence for one gene; this is known as monocistronic.
There is an overlap between transcription and translation, with protein synthesis beginning during transcription. This is known as coupled transcription–translation. This can occur because the DNA and ribosomes are in the cytosol together.	There is no overlap between transcription and translation. The transcription of DNA occurs in the nucleus, and translation and protein synthesis occur in the cytoplasm.
Prokaryotes mostly contain exons, with very few introns	Eukaryotes have introns and exons.
Because prokaryotes contain few introns, RNA processing is rarely required after transcription. RNA processing is much simpler than in eukaryotic cells.	RNA processing removes the introns (non-coding regions) so only the exons (coding regions) are translated.
Prokaryotes have about three different initiation factors.	Eukaryotes have around 10 different initiation factors.
No 5' cap is added to mRNA.	A methylguanosine triphosphate molecule, called a 5' cap, is added to the 5' end of the mRNA.
No poly(A) tail is added to mRNA.	A poly(A) tail is added to the 3' end of mRNA.

AMINO ACIDS, POLYPEPTIDES AND PROTEINS

Protein molecules are more complex than other **biomolecules** like carbohydrates or lipids. For example, the haemoglobin that carries oxygen in human blood has a chemical formula of $C_{2952}H_{4664}O_{832}N_{812}S_8Fe_4$. Proteins make up more than 50% of the dry weight of cells. There are thousands of different kinds of proteins, their functions vary widely and they are essential to the wellbeing of any organism. Although carbohydrates and lipids are similar in all plants and animals, organisms can have a variety of unique proteins that are specific to a species. This section explains the way proteins are synthesised inside cells from the inherited DNA code. Section 4.3 will discuss proteins in more detail.

Proteins are **biomacromolecules** made of chains of subunits called amino acids. Amino acids are linked by a chemical bond called a **peptide bond** and form polypeptides or polypeptide chains (Figure 4.2.16). Polypeptide means 'many peptide bonds'. A **peptide** is a linear sequence of fewer than 50 amino acids. A **dipeptide** is a molecule consisting of two amino acids joined with a peptide bond. A polypeptide is a molecule consisting of many (more than 50) amino acids joined together by peptide bonds. A protein is formed by one or more polypeptides arranged in a biologically functional way. In other words, 'protein' is the term used for a fully functioning molecule while 'polypeptide' refers to a non-functioning component.

There are 20 different amino acids commonly found in proteins. Nine of these are known as essential amino acids because they cannot be produced by humans. Humans must obtain these from their food. The properties of many proteins are determined by their shape, which is determined in turn by their amino acid sequence.

BIOFILE CCT PSC

Amino acids in the human diet

In the human diet, amino acids can be classified into three main groups.

- Essential amino acids: the body cannot synthesise these, we must obtain them from our diet (Figure 4.2.15). The nine essential amino acids are histidine, isoleucine, leucine, lysine, methionine, phenylalanine, threonine, tryptophan and valine.
- Non-essential amino acids: the body can produce these if not obtained from the diet. The five non-essential amino acids include alanine, asparagine, aspartic acid, glutamic acid and serine.
- Conditional amino acids: the body only requires these in times of illness or stress. The conditional amino acids include arginine, cysteine, glycine, glutamine, proline and tyrosine.

FIGURE 4.2.15 A collection of foods and supplements high in amino acids

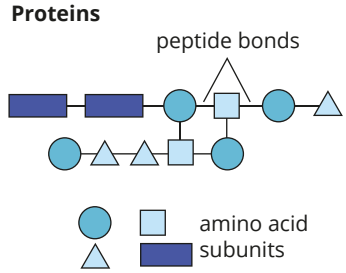

FIGURE 4.2.16 The structure and bonding of proteins. Amino acid monomer units are bonded into peptides, then into polypeptides, then structured into functional proteins.

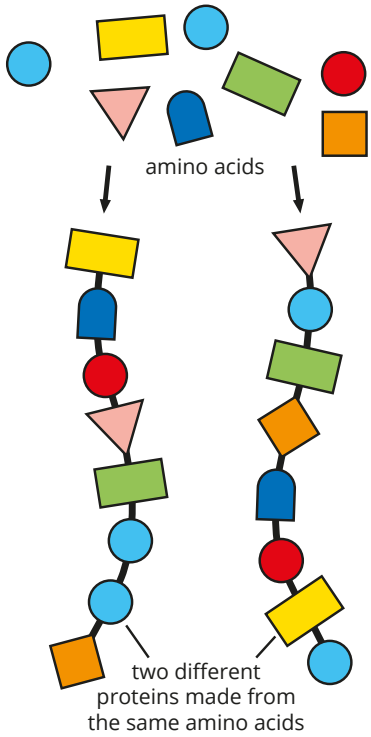

FIGURE 4.2.17 The diagram models how the same amino acids can be sequenced in a different order to make different polypeptide chains that, in turn, will form different proteins.

There are many steps involved in producing a functional protein. Although protein structure, size and function are quite diverse, all proteins are made up of amino acids. These smaller subunits (or monomers) are joined together in a particular order to form polypeptide chains (Figure 4.2.17). The polypeptide chains are then folded and coiled into proteins (Figure 4.2.18). Section 4.3 will explain the structure of proteins in greater detail.

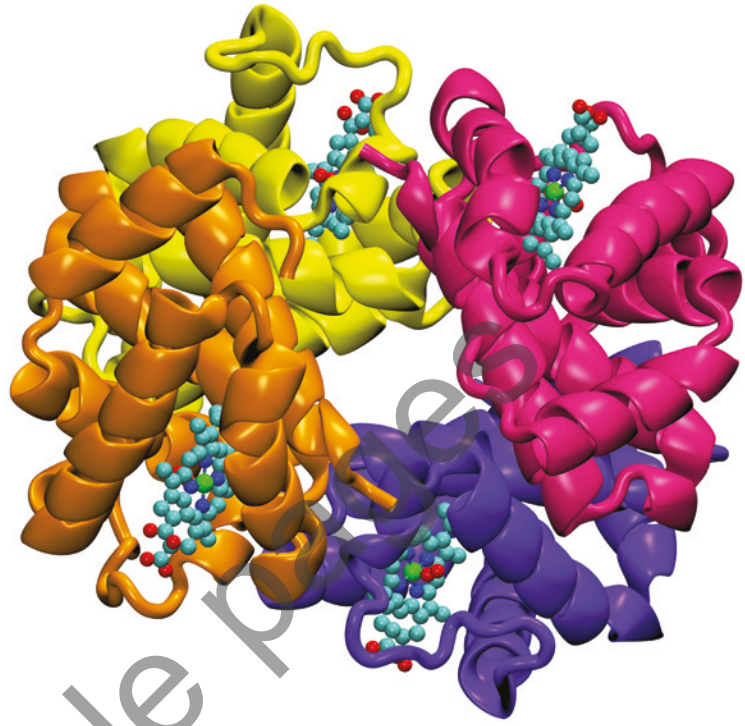

FIGURE 4.2.18 Haemoglobin is made up of four polypeptide subunits (coloured ribbon structures). Each has a haem group (turquoise), which is the oxygen-binding site (O_2=paired red spheres). Within each haem group there is one atom of iron (green). The chemical formula for an Hb molecule is $C_{2952}H_{4664}O_{832}N_{812}S_8Fe_4$.

BIOLOGY IN ACTION CCT ICT

Protein forensics and LOC

Because many proteins are unique to an organism, scientists can test a protein sample to determine which animal it came from. Some shops and restaurants have been caught selling shark as barramundi or putting horsemeat into hamburgers. The suspect protein sample can be analysed to determine its origin.

A technology referred to as lab-on-a-chip (LOC) miniaturises and compacts the different test processes that a researcher or a diagnostic lab technician uses. A tiny fluid sample is added to one end of the microchip-sized wafer of glass or plastic and the molecules of the suspect protein are channelled past circuits of nanometre-sized chemical and physical tests (Figure 4.2.19). The device is still being perfected and holds great promise for testing samples in the field or when the sample is very small in volume.

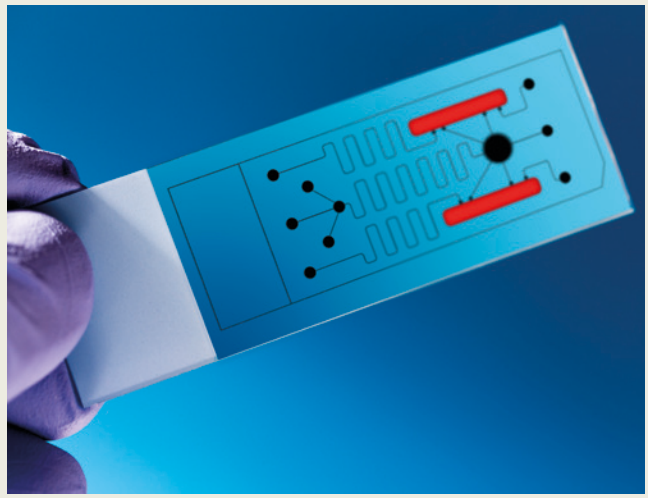

FIGURE 4.2.19 Biochip for identifying proteins using the LOC technology. Test systems for analysis of a sample are integrated into one microchip-sized thin glass plate.

The function and importance of polypeptide synthesis

The genome consists of many thousands of genes. A cell can express a selection of these genes at a given time. The genes that are expressed determine which proteins are produced, giving the cell its functionality and characteristics. Gene expression is the process through which information from a gene is used to synthesise a specific functional gene product—a polypeptide or length of RNA. Gene expression in eukaryotes is tightly regulated by multiple mechanisms at different points, usually in the transcription phase. The regulation determines that the correct polypeptides and proteins are produced when and where they are required. For example:

- growth hormone proteins are produced in infants, children and teenagers but switched off at other times
- plasma B cells of the immune system are regulated to only produce antibody proteins when they are required to fight an infection
- enzyme proteins are recycled but new ones will be synthesised to replace those that have reached the end of their useful life
- continual production of haemoglobin in the bone marrow of vertebrates.

BIOFILE L

Gene names

A gene name is always italicised, to distinguish it from the protein it encodes. For example, the disorder phenylketonuria (PKU) is caused by a mutation in the *PAH* gene that normally expresses the enzyme phenylalanine hydroxylase (PAH). The inherited disease of cystic fibrosis (CF) comes from mutation of the *CFTR* gene, which codes for CFTR protein, which regulates the movement of chloride ions across cell membranes.

BIOFILE CCT

Haemoglobin

Haemoglobin (Hb) is the oxygen transport protein carried on red blood cells of vertebrates. In humans, a functional haemoglobin protein molecule is built from four polypeptide chains. The polypeptides are constantly being synthesised by cells in the bone marrow so that newly differentiated red blood cells will carry haemoglobin (Figure 4.2.20).

Mature red blood cells have no nucleus and therefore cannot form new proteins while they circulate in the blood. This limits their lifetime to around 120 days, and means it is necessary for constant production of replacement red blood cells by the bone marrow—estimated at the extraordinary rate of around two million new red blood cells per second. Haemoglobin has the critical function of carrying oxygen to all parts of the body so that cells can produce energy using cellular respiration. Therefore, it is essential that gene expression in bone marrow cells is regulated to produce a constant supply of polypeptides for constructing the protein structure of haemoglobin.

When you donate blood, you lose red blood cells. Special cells in the kidneys, called peritubular cells, sense the decreased level of oxygen in the blood and start secreting a protein called erythropoietin. It passes through the bloodstream until it reaches the bone marrow where the erythropoietin triggers stem cells to develop into more red blood cells, rather than white blood cells or platelets.

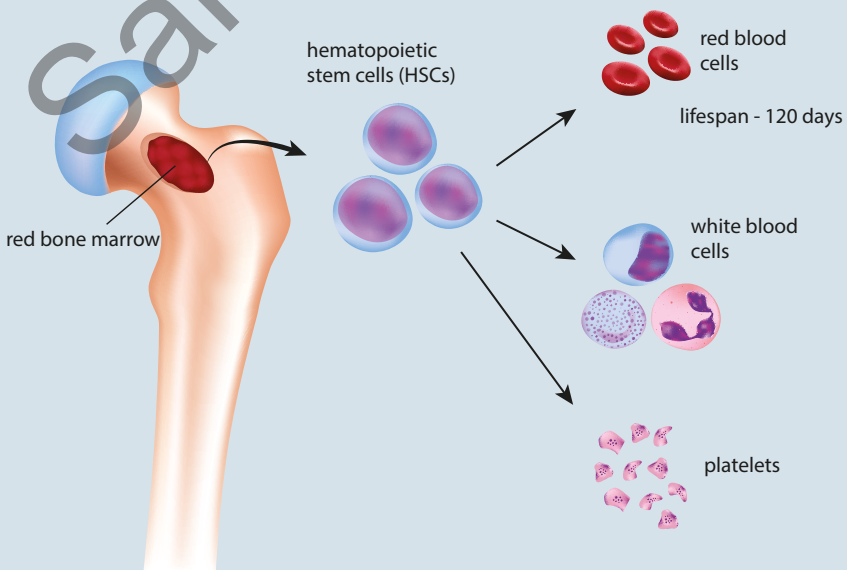

FIGURE 4.2.20 Blood cell formation from bone marrow. Haemoglobin protein must be available for red blood cells to fulfil the vital function of oxygen transport.

GENOTYPE, PHENOTYPE AND THE ENVIRONMENT

An organism's phenotype is all its observable characteristics. It is the result of inheritance and the effects of the organism's environment. The genotype is the set of alleles present in the DNA of an individual organism. The genotype is the result of inheritance and any mutations in the germ cells.

The **proteome** is the complete set of proteins expressed by the genome (the complete set of genes) of an individual cell or organism at a given time. The proteome varies between cell type, developmental stage and environmental conditions. Although a cell may contain the entire genome, only specific genes will be expressed, or 'switched on', at any given time. This ensures a cell produces only the proteins required for the specific functions it carries out.

The **phenotype** is the set of characteristics expressed from the genetic information of a cell. That is, the part of the **genotype** that has been switched on to allow transcription and translation to produce polypeptides, then proteins.

- An organism's genotype and environment determine its phenotype.
- If an individual with a given genotype develops in one environment, its phenotype may be different than if it had developed in a different environment.
  - For example, the average height of humans has gradually increased in the last few hundred years because of improved nutrition.
  - On the other hand, a human cannot grow to the full height potential that is coded in their genotype if they are severely undernourished as a child or if a bone disease affects their growth.
  - A tree will be stunted in height if it does not receive the water and minerals needed to reach its genetic potential or if it grows with continual strong wind, in constant shade or is grown in a small pot where it becomes root-bound.
  - A human's skin colour depends partly on the amount of sunlight exposure. In the short term, it can be altered by air temperature or exercise, such as being red-faced after strenuous exercise or pale-skinned on a very cold day.

Other examples of environmental influence on phenotype are:

- the development and severity of inherited disorders such as phenylketonuria (PKU)
- flower colour variation with soil pH
- seasonal variation in the fur colour of Arctic foxes
- feather colour variation in flamingos with different levels of carotenoids (organic pigments produced by algae, bacteria and fungi) in their diet (Figure 4.2.21).

You will learn more about the effect of the environment on phenotype in Chapter 5.

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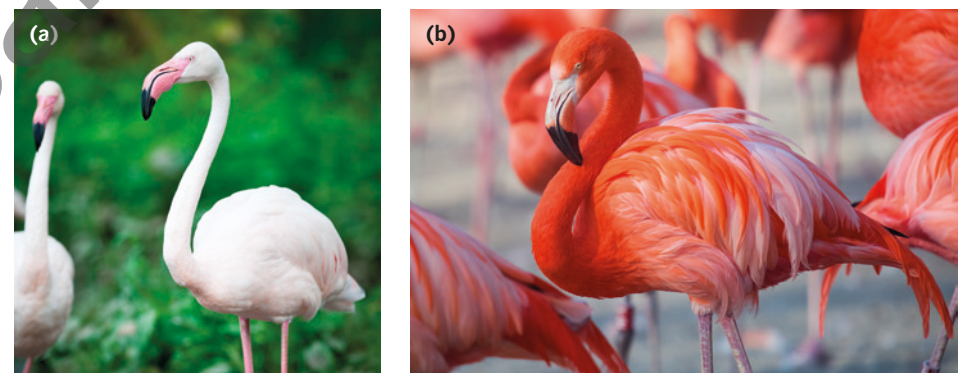

FIGURE 4.2.21 The colour of a flamingo's feathers (phenotype) is determined by their dietary intake of carotenoids (environment). Carotenoids are pigments produced by some species of algae, bacteria and fungi. The flamingos' diet of brine shrimp and blue-green algae is a rich source of carotenoids. Feather colour varies from (a) light pink to (b) dark pink/red. The higher the intake of carotenoids, the pinker the flamingo's feathers will be.

4.2 Review

SUMMARY

- DNA stores hereditary information, carrying the instructions that code for the production of proteins and functional RNA molecules, in a specific sequence of nucleotides.
- The genetic code is the set of rules about how the instructions carried in nucleic acids are translated to synthesise proteins and functional RNA molecules. In DNA this information is stored as a three-letter code of nucleotides known as a triplet. When these triplets are transcribed into mature mRNA, they are then known as codons. On tRNA the group of three bases is called an anticodon because these bases are the opposite partners (complements) of the mRNA codons.
- The genetic code is universal and degenerate. There are 64 possible codons of three nucleotides each (e.g. UAC) for the 20 amino acids.
- A gene is a region of DNA that codes for a polypeptide in both prokaryotes and eukaryotes. Unique to eukaryotes, a gene may also code for a functional RNA molecule.
- Eukaryotic genes have many structural features in common:
  - Stop and start instructions—indicate where transcription of mRNA starts and stops. Stop codons do not code for amino acids.
  - Promoter region—the site at which the RNA polymerase attaches to the gene to begin transcription (sometimes called the TATA box).
  - Exons—coding DNA regions.
  - Introns—non-coding DNA regions.
- Gene expression is the process in which the information stored in a gene is used to synthesise a functional gene product (polypeptide or RNA). Gene expression is regulated so that it occurs if the particular protein or RNA is required by the cell.
- RNA is a short, usually single-stranded, nucleic acid.
- RNA contains nucleotides that are made up of ribose sugar, a phosphate and one of four nitrogenous bases (adenine, cytosine, guanine and uracil).
- The role of RNA is to express the information contained in the nucleotide sequence of a gene to synthesise polypeptides.
  - mRNA is produced by a process called transcription in the nucleus—a single-stranded nucleic acid that carries a copy of the genetic sequence in DNA, specifying the amino acid sequence for a polypeptide.
  - rRNA makes up part of a ribosome. Ribosomes are the sites where the information in the mRNA is translated into a chain of amino acids.
  - tRNAs carry specific amino acids to ribosomes to form polypeptide chains by the process called translation.
- Polypeptide synthesis in eukaryotes occurs in three stages: transcription, RNA processing and translation.
- Transcription occurs in the nucleus and involves RNA polymerase transcribing the DNA into a primary RNA transcript.
- The primary RNA transcript is spliced to remove the introns, and sometimes some exons, resulting in mature mRNA. The mature mRNA then leaves the nucleus.
- Translation occurs on a ribosome in the cytoplasm. The codons on mRNA are translated into a sequence of amino acids to form a polypeptide chain. The amino acids are delivered individually by their specific tRNA molecules each with an anticodon that complements the codon of the mRNA.
- After synthesis, polypeptide chains are folded and combined to form fully functional proteins. 'Protein' is the term used for a fully functioning molecule while 'polypeptide' refers to a non-functioning component.
- Polypeptide synthesis and protein formation is crucial to the functioning of all cells. Proteins are important because enzyme proteins control cell metabolism; other proteins are hormones and antibodies; proteins are important receptor, structural, storage and transport molecules; and they are important in movement.
- Changes in the amino acid sequence can lead to faults in proteins and cause health issues.
- The genotype plays an important role in determining the phenotype but gene expression can be affected by environmental influences such as temperature, soil pH, nutrition, availability of key chemicals and sunlight exposure.

4.2 Review *continued*

KEY QUESTIONS

1 Distinguish between peptides, dipeptides and polypeptides, using a diagram to support your answer.

2 Explain how RNA differs from DNA, mentioning at least three features that differentiate them.

3 Name the three types of RNA and outline their basic functions.

4 Identify the three stages of polypeptide synthesis in eukaryotes.

5 Transcription occurs in three stages: initiation, elongation and termination. Rearrange the table to match the transcription event with the correct stage at which it occurs.

Stage of transcription	Transcription event
initiation	• The RNA polymerase moves along the DNA molecule, producing a strand of mRNA. • The RNA polymerase detaches, releasing the mRNA and allowing the DNA molecule to reform.
elongation	• RNA polymerase uses a strand of DNA as a template, attaching nucleotides (A, U, G, C) by complementary base pairing. • Transcription factors combine with the region at the start of the gene, known as the promoter. • RNA polymerase reaches the termination site of the gene (stop codon) and translation ends.
termination	• RNA polymerase attaches to the promoter, unwinding and unzipping the DNA molecule by breaking the weak hydrogen bonds between the two strands to expose the bases.

6 Like transcription, translation occurs in three stages: initiation, elongation and termination. However, the events during each of these stages are different. Rearrange the table to match the translation event with the correct stage at which it occurs.

Stage of translation	Translation event
initiation	• Following the attachment of the amino acid methionine, another tRNA, with a complementary anticodon to the next codon on the mRNA, attaches and adds its specific amino acid to the growing polypeptide chain. • The tRNA reaches a stop codon.
elongation	• A small ribosomal subunit attaches to the 5' end of an mRNA strand. It then moves along the mRNA until it reaches a start codon (AUG). • The polypeptide chain is released from the ribosome into the cytoplasm or the endoplasmic reticulum.
termination	• The ribosome then releases the tRNA and moves further along the mRNA strand. At each codon, a new tRNA binds and adds another amino acid. • A tRNA molecule with an anticodon (UAC) brings the amino acid methionine to the mRNA. The tRNA molecule joins to the mRNA start codon, attaching by complementary base pairing between the codon and anticodon.

7 Outline the main structural features of eukaryotic genes and their functions.

8 Define what is meant by 'universal genetic code'.

9 There is a longstanding debate referred to as nature versus nurture. In the debate, nature refers to inherited information and ~~nature~~ refers to environmental effects on phenotype. Use your scientific knowledge to assess how genes and the environment both influence the phenotype of an organism.
Support your answer with examples.